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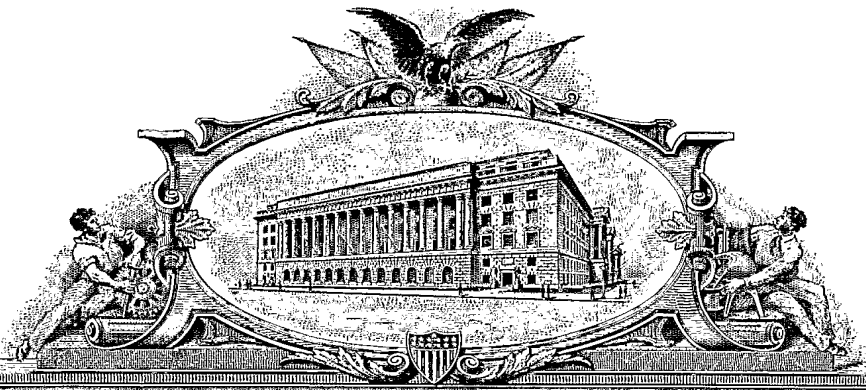
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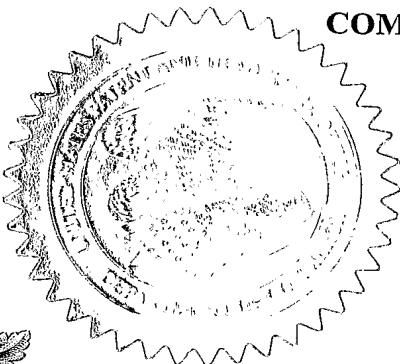
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PROVISIONAL APPLICATION FOR PATENT COVER SHEET This is a request for filing a PROVISIONAL APPLICATION FOR PATENT under 37 CFR 1.53(c).

INVENTOR(S)					
Given Name (first and middle [if any])		Family Name or Surname		Residence (City and either State or Foreign Country)	
Antonio		FERRANTE		Queensland, Australia	
<input type="checkbox"/> Additional inventors are being named on the _____ separately numbered sheets attached hereto					
TITLE OF THE INVENTION (280 characters max)					
THERAPEUTIC AND CARRIER MOLECULES					
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ENCLOSED APPLICATION PARTS (check all that apply)					
<input checked="" type="checkbox"/> Specification Number of Pages 215		<input type="checkbox"/> CD(s), Number 			
<input checked="" type="checkbox"/> Drawing(s) Number of Sheets 13		<input type="checkbox"/> Other (specify) 			
<input type="checkbox"/> Application Data Sheet. See 37 CFR 1.76					
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Respectfully submitted,

SIGNATURE

TYPED or PRINTED NAME

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516-742-4343

Date

01/30/04

REGISTRATION NO.

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Docket Number:

P17418

USE ONLY FOR FILING A PROVISIONAL APPLICATION FOR PATENT

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Invention:

THERAPEUTIC AND CARRIER MOLECULES

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THERAPEUTIC AND CARRIER MOLECULES

BACKGROUND OF THE INVENTION

5 FIELD OF THE INVENTION

The present invention relates generally to compounds comprising a hydrocarbon chain portion and chemical derivatizations of the chain to produce useful therapeutic and prophylactic molecules. As well, the hydrocarbon chain portion is a carrier molecule for
10 functional groups or agents. The compounds of the present invention are particularly useful in the treatment and prophylaxis of a range of conditions including cancers, protein kinase c(PKC)- or NFκB-related or associated conditions, cardiovascular conditions, pain, inflammatory conditions, vascular or immunological conditions such as diabetes, neurological conditions and infection by a range of viruses or prokaryotic or eukaryotic
15 organisms.

DESCRIPTION OF THE PRIOR ART

Bibliographic details of references in the subject specification are also listed at the end of
20 the specification.

Reference to any prior art in this specification is not, and should not be taken as, an acknowledgment or any form of suggestion that that prior art forms part of the common general knowledge in any country.
25

Fatty acids are one of the most extensively studied classes of compounds due to their important role in biological systems (Ferrante *et al.*, In *The Neutrophils: New outlook for the old cells* [Ed Garbilovich] Imperial College Press 4:79-150, 1999; Sinclair and Gibson (Eds) Invited papers from the Third International Congress, American Oil Chemists' Society, Champaign, Illinois, 1-482, 1992). Fatty acids consist of saturated, monosaturated
30 and polyunsaturated fatty acids having a chain length from 4 to 30 carbon atoms.

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Polyunsaturated fatty acids (PUFAs) contain 16 to 30 carbon atoms with two or more methylene-interrupted *cis*-double bonds.

5 PUFA nomenclature includes recitation of the number of carbon atoms in the hydrocarbon chain, the number of double bonds and the position of the first double bond from the terminal methyl group (the ω -carbon atom). For example, the PUFA arachidonic acid contains 20 carbon atoms and four methylene-interrupted *cis*-double bonds commencing six carbons from the ω -carbon, *viz*: this PUFA is referred to as "arachidonic acid (20:6 n-6)".

10 PUFAs can be divided into four families based on the fatty acids from which they are derived: linoleic acid (18:2 n-6), α -linolenic acid (18:3 n-3), oleic acid (18:1 n-9) and palmitoleic acid (16:1 n-7). The n-6 and n-3 PUFAs cannot be synthesized by mammals and are known as essential fatty acids (EFAs). They are acquired by mammalian bodies indirectly through desaturation or elongation of linoleic and α -linolenic acids, which must
15 be supplied in the diet.

It is now well appreciated that ω -3 fatty acids confer some protection against a range of diseases. Synthetic fats have been synthesized which are useful in the treatment of a variety of conditions.

20

International Patent Publication Nos. WO 96/11908, WO 96/13507, WO 97/38688, WO 01/21172 and WO 01/21575 describe a range of PUFAs referred to as the MP Series, PT Series LX Series and MP-PT hybrid-series. Some of these PUFAs, such as those of the MP Series, have reduced susceptibility to breakdown and hence are far less likely to cause
25 the production of oxygen radicals which is the consequence of the metabolism of the natural ω -3 fatty acids. PT PUFAs also have this property of resisting breakdown but in addition are more soluble. MP-PT hybrids are particularly useful anti-inflammatory agents.

30 As indicated above, naturally occurring ω -3 fatty acids have found to be useful in treating a range of conditions including rheumatoid arthritis, multiple sclerosis, inflammatory

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bowel disease and systemic lupus. The PUFAs of the MP, PT, LX and MP-PT hybrid families have also been proposed for the treatment of malaria, to stimulate or inhibit neutrophil activity, to treat T-cell diseases and in the treatment of cancer.

- 5 There is a need to determine the full range of activities of the PUFAs and to identify naturally occurring or to generate synthetic derivatives which have therapeutic potential.

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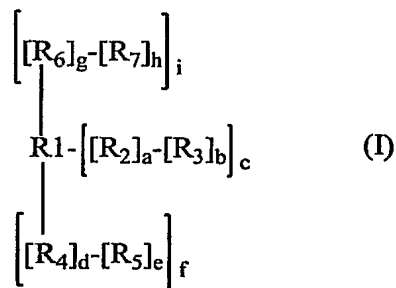
SUMMARY OF THE INVENTION

Throughout this specification, unless the context requires otherwise, the word "comprise", or variations such as "comprises" or "comprising", will be understood to imply the
 5 inclusion of a stated element or integer or group of elements or integers but not the exclusion of any other element or integer or group of elements or integers.

In accordance with the present invention, it is proposed that the PUFAs are useful in the treatment *inter alia* of conditions associated with or involving protein kinase C β (PKC β)
 10 and/or NF κ B and in the treatment of pain, inflammation, vascular or immunological conditions such as diabetes, cardiovascular conditions, atherosclerosis, neurological conditions and infection by a range of viruses, prokaryotes or eukaryotes.

In particular, the present invention contemplates a method for the treatment or prophylaxis
 15 of a condition selected from a NF κ B related or associated condition, a PKC β related or associated condition, vascular or immunological conditions such as diabetes, inflammation, neurological conditions, cardiovascular disease and pain in a subject said method comprising administering to said subject an effective amount of a compound having the structure of Formula (I):

20



wherein

25 R_1 is a saturated or unsaturated hydrocarbon chain of from about 9 to about 26 carbon atoms and which is optionally carries one or more of a oxa, thia, hydroxy, hydroperoxy,

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epoxy and peroxy substitution;

each of R₂, R₄ and R₆ is selected from O₂, NO, NO₂, S(O)_x, C(H)_y, H, COOH, P(X)_δ(Y),

5 N(H)_z, C=O, OH, $\text{---}\overset{\text{O}}{\parallel}\text{C}\text{---}\text{NH}\text{---}$, C₁₋₆ alkyl, C₁₋₆ alkoxy, amino, mono-acid di-C₁₋₆ alkylamino, C₁₋₆ alkylthio, S(O)_x-C₁₋₃ alkyl, C₁₋₆ alkoxy carbonyl, halo selected from fluoro, chloro, bromo and iodo, oxo, amidino and guanidino, C₂₋₁₂ alkenyl, C₂₋₁₂ alkynyl, aryl, heteroaryl and cyano, wherein x and z are 0, 1 or 2 and y is 0, 1, 2 or 3 and X is O, S or NR₈, Y is OR₉, SR₁₀ or NR₁₁R₁₂ and R₈, R₉, R₁₀, R₁₁ and R₁₂ are selected from H, alkyl, alkenyl, alkynyl, aryl and heteroaryl, δ is 0 or 1;

10

each of R₃, R₅ and R₇ is respectively [(CH₂)_j (COOH)_k]_l, [(CH₂)_m (COOH)_n]_o and [(CH₂)_p (COOH)_q]_r, wherein each of j, m and p is 0, 1, 2, 3, 4, 5 or 6, each of k, n and q is 0, 1 or 2, and each of l, o and r is 0 or 1,

15 each of c i and f is 0 or 1 or 2;

each of a, d and g is 0 or 1 or 2;

each of b, e and h is 0 or 1 or 2;

20

said administration being for a time and under conditions sufficient to prevent the condition or to ameliorate one or more symptoms of the condition.

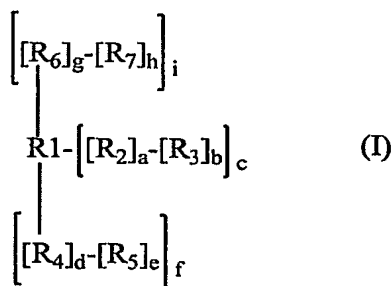
25 The present invention extends to isolated naturally occurring PUFAs as well as synthetic, modified molecules. The subject molecules also include a range of hybrids in which the PUFA is conjugated to an L- or D-amino acid or a chemical analog of an amino acid.

30 The present invention further extends to compounds of general Formula (I) as defined above in isolated form or in a composition such as a pharmaceutical composition or formulation.

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The present invention further provides for the use of a compound of general Formula (I) as defined above in the manufacture of a medicament for the treatment of a condition selected from a condition associated with or involving NFκB, PKCβ, pain, vascular or immunological conditions such as diabetes and cardiovascular disease, atherosclerosis, neurological conditions, inflammation and infection by a range of viruses, prokaryotes and eukaryotes.

The present invention also provides a compound of Formula (I):



wherein

15

R₁ is a saturated or unsaturated hydrocarbon chain of from about 9 to about 26 carbon atoms and which is optionally carries one or more of a oxa, thia, hydroxy, hydroperoxy, epoxy and peroxy substitution;

20 each of R₂, R₄ and R₆ is selected from O₂, NO, NO₂, S(O)_x, C(H)_y, H, COOH, P(X)_δ(Y),

N(H)_z, C=O, OH, $\text{---}\overset{\text{O}}{\parallel}\text{C---NH---}$, C₁₋₆ alkyl, C₁₋₆ alkoxy, amino, mono-acid di-C₁₋₆ alkylamino, C₁₋₆ alkylthio, S(O)_x-C₁₋₃ alkyl, C₁₋₆ alkoxy carbonyl, halo selected from fluoro, chloro, bromo and iodo, oxo, amidino and guanidino, C₂₋₁₂ alkenyl, C₂₋₁₂ alkynyl, aryl, heteroaryl and cyano, wherein x and z are 0, 1 or 2 and y is 0, 1, 2 or 3 and X is O, S or NR₈, Y is OR₉, SR₁₀ or NR₁₁R₁₂ and R₈, R₉, R₁₀, R₁₁ and R₁₂ are selected from H, alkyl,

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alkenyl, alkynyl, aryl and heteroaryl, δ is 0 or 1;

each of R_3 , R_5 and R_7 is respectively $[(CH_2)_j (COOH)_k]_l$, $[(CH_2)_m (COOH)_n]_o$ and $[(CH_2)_p (COOH)_q]_r$, wherein each of j , m and p is 0, 1, 2, 3, 4, 5 or 6, each of k , n and q is 0, 1 or 2,
5 and each of l , o and r is 0 or 1,

each of c , i and f is 0 or 1 or 2; and

each of a , d and g is 0 or 1 or 2;

10

each of b , e and h is 0 or 1 or 2.

BRIEF DESCRIPTION OF THE FIGURES

Figure 1 is a diagrammatic representation showing the principle mechanism involving T-lymphocytes, leukocytes, macrophages and other cells of the immune system.

5

Figure 2 is a diagrammatic representation of a structure of MP3 (β -oxa-23:4n-6).

Figure 3 is a diagrammatic representation showing the suppression of TNF-stimulated endothelial cell adhesion molecule expression by cells were pretreated with MP3 (1h) before being stimulated with TNF for the times indicated. Adhesion molecule expression was determined by ELISA.

10

Figure 4 is a diagrammatic representation showing the suppression of LPS-stimulated leukocyte infiltration into the peritoneal cavity (a) and suppression of E-selectin expression by aortic endothelium (b) by MP3.

15

Figure 5 is a diagrammatic representation showing the prevention of TNF-stimulated loss of I κ B α in HUVEC by MP3 or 22:6n-3 cells were pretreated with MP3 or 22:6n-3 (1 h), stimulated with TNF (15 min) lysed and the lysate subjected to Western blot analysis using anti-I κ B α antibody.

20

Figure 6 is a diagrammatic representation showing the suppression of PKC β 1 translocation in glucose-stimulated mesangial cells (a) and in the glomeruli of a diabetic rat (b). Mesangial cells were pretreated with MP5 or vehicle (ethanol) for 1 hour before being incubated with 25 mM glucose for 5 days. Male rats were rendered diabetic with streptozotocin and MP5 or vehicle (ethanol) was administered for 7 days after confirmation of diabetes. The cells and glomeruli were sonicated and particulate fraction-associated PKC β 1 was determined by Western blot analysis. High glucose and diabetes increased PKC β 1 in the particulate fraction. MP5 inhibited this effect.

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Figure 7 is a representation showing comparison of the ability of MP3 (β -oxa-23:4n-6) PMA (100nmol/l) and 22:6n-3 to stimulate the neutrophil respiratory burst. Neutrophils were treated with DPC (Control), 23:4n-6, PMA or 22:6n-3 and then tested for chemiluminescence activity. The fatty acids were used at 20 μ mol/l. The results are the mean \pm SEM of quadruplicates and is representative of two other experimental runs.

Figure 8 is a representation showing effect of β -oxa, β -thia and natural PUFA on TNF-enhanced neutrophil adherence to HUVEC. HUVEC were pretreated with the fatty acids (20 μ mol/l) for 60 min at 37°C before being stimulated with TNF (125U/200 μ l medium) for 4h at 37°C. The cells were then co-incubated with neutrophils (5×10^5 cells/well) at 37°C for 30 min and the degree of neutrophil adherence quantitated. The results are expressed as % of control and represent the mean \pm SEM of three separate experiments each performed in triplicate. * p < 0.05, *** p < 0.001, for significant differences between pre-treatment with fatty acid and control (one-way analysis of variance followed by the Dunnett test for multiple comparisons).

Figure 9 is a representation showing effect of MP3 derivatives on TNF-enhanced neutrophil adherence to HUVEC. HUVEC were pre-treated with MP3 (20 μ mol/l), β -oxa-23:4n-6 derivatives (20 μ mol/l) or diluent (control) for 60 min and then challenged with TNF (125 U/ 200 μ l medium) for a further 4 h. The ability of HUVEC to adhere neutrophils was then assessed. The results are expressed as % of control and represent the mean \pm SEM of three separate experiments each performed in triplicate. *** p < 0.001, for significant differences between pre-treatment with MP3 (β -oxa-23:4n-6) or derivative and control (one-way analysis of variance followed by the Dunnett test for multiple comparisons). Abbreviations used: β -oxa-23:4n-6ME, β -oxa-23:4n-6 methyl ester; β -oxa-23:0, saturated form of β -oxa-23:4n-6; β -oxa-23:4n-6OH, 18-monohydroxy- β -oxa-23:4n-6; β -oxa-23:4n-6OOH, 18-monohydroperoxy- β -oxa-23:4n-6.

Figure 10 is a representation showing effect of MP3 (β -oxa-23:4n-6) and 20:4n-6 on time-related changes in TNF- α -induced E-selectin, ICAM-1 and VCAM-1 expression on

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HUVEC. HUVEC were pre-treated with 20 $\mu\text{mol/l}$ β -oxa-23:4n-6 (closed triangles), 20 $\mu\text{mol/l}$ 20:4n-6 (open squares), or DPC (control) for 60 min and then further incubated with TNF- α (125 U/200 μl medium) for up to 24 h. The expression of E-selectin, ICAM-1 and VCAM-1 adhesion molecules was determined by ELISA. The results are expressed as
 5 % of control and represent the mean \pm SEM of three separate experiments each performed in triplicate. * $p < 0.05$, ** $p < 0.01$, *** $p < 0.001$, for significant differences between pre-treatment with fatty acid and corresponding control at a particular time point (one-way analysis of variance followed by the Dunnett test for multiple comparisons). **Inset:** The effect of β -oxa-23:4n-6 on TNF- α -induced expression of E-selectin mRNA in HUVEC.
 10 HUVEC were pre-incubated with β -oxa-23:4n-6 (20 $\mu\text{mol/l}$) or DPC (control) in 1 ml of medium at 37°C for 60 min. After the addition of TNF- α , the cells were further incubated at 37°C for 2 h. E-selectin mRNA expression was then determined and the results expressed as relative %. Results are the mean \pm SEM of three separate experiments each performed in quadruplicate. * $p < 0.0001$, for significant differences between pre-treatment
 15 with β -oxa-23:4n-6, and control (two-tailed Student's t-test for unpaired data).

Figure 11 is a representation showing (A) Effect of MP3 on *in vivo* inflammatory response measured as delayed type hypersensitivity (DTH) to sheep erythrocytes and LPS-induced influx of neutrophils and mononuclear cells in the peritoneal cavity in BALB/c mice. In
 20 the DTH experiments mice were injected with sheep erythrocytes subcutaneously, challenged with the antigen in the hind foot pad six days later and the amount of foot pad swelling measured 48h later. One hour prior to challenge mice were given 10mg/kg body weight of β -oxa fatty acid in 7% w/v DMSO as vehicle intraperitoneally. For the peritoneal cavity inflammation, mice were given intravenously 40mg/kg MP3
 25 intravenously and 6h later injected with LPS intraperitoneally. The cellular infiltrates were examined 24 and 72h later. The data, expressed as % of control, are presented as mean \pm SEM of 10 and 5 mice for DTH and peritoneal inflammation, respectively. Analysis of data by two-tailed student's t-test: ** $p < 0.01$, *** $p < 0.001$. (B) Shows the effect of β -oxa-23:4n-6 on LPS-induced expression of E-selectin in aortic endothelium of BALB/C mice.
 30 Mice were treated intravenously with the fatty acid and 2h later injected intraperitoneally

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with LPS. After 5 h the aortas were isolated, cut into small pieces and incubated with a monoclonal antibody to mouse E-selection (or isotype matched control) (Becton Dickinson, California) followed by an HRP-conjugated secondary antibody and then with the substrate ABTS (ELISA method). The data, expressed as % of control, are presented as
 5 mean \pm SEM of ten mice per group and is representative of two experimental runs. Analysis of the data by the two-tailed student's t-test: ** $p < 0.01$.

Figure 12 is a representation showing the chemical structure of MP3 (β -oxa-23:4n-6) and of the monohydroxylated derivatives of β -oxa-23:4n-6 formed *via* the lipoxygenase
 10 pathway in HUVECs (15-monohydroperoxy- β -oxa-23:4n-6 was the predominant product).

Figure 13 is a representation showing the effects of lipoxygenase/cyclooxygenase inhibitors and antioxidants on the modulation of E-selectin expression on HUVEC by β -
 15 oxa-23:4n-6. HUVEC were pretreated with NDGA, baicalein, MK886, indomethacin, Vitamin E, or diluent (control) for 15 min. The cells were then further incubated with 20 μ mol/l β -oxa-23:4n-6 or diluent (control) for 60 min followed by TNF- α (125U/200 μ l medium) for 4 h and the expression of E-selectin adhesion molecule was determined. The results are expressed as % inhibition of the suppressive effect of β -oxa-23:4n-6 and
 20 represent the mean \pm SEM of three separate experiments each performed in quadruplicate. * $p < 0.01$, for significant differences between pretreatment with inhibitor and corresponding control (one-way analysis of variance followed by the Dunnett test for multiple comparisons).

Figure 14 is a representation showing the effect of MP3 (β -oxa 23:4n-6) and DHA on TNF-induced degradation of I κ B α in HUVEC. Cells were pretreated with the fatty acids (20 μ mol/l) for 30 min and then stimulated with TNF (125 U/ml) for 10 min. After cell
 lysis the proteins were analysed by western blots using anti-I κ B α antibodies. **(B)** The effects of β -oxa-23:4n-6 on TNF-induced activation of transcriptional factor, NF κ B in
 30 HUVEC. Cells were pretreated with β -oxa-23:4n-6 (20 μ mol/l) for 30 min and then

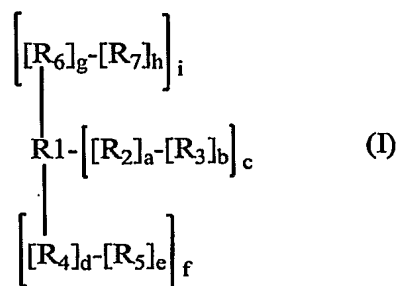
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stimulated with TNF for 2 h. After cell lysis, nuclear fractions were prepared, nuclear proteins separated by SDS PAGE (12% w/v gel), transferred to nitrocellulose and probed with an anti-NF κ B p65 antibody (Santa Cruz). Densitometric analysis of data from 3 experiments showed that β -oxa 23:4n-6 reduced TNF-stimulated nuclear accumulation
5 of NF κ B by $66 \pm 2\%$ (mean \pm SEM) ($p < 0.001$, two-tailed student's t-test). (c) The effect of β -oxa 23:4n-6 on TNF-stimulated activation of IKK. Cells were pretreated with β -oxa 23:4n-6 (20 μ mol/l) for 30 min and then stimulated with TNF for 5min. After cell lysis IKK was immunoprecipitated with anti-IKK α antibody and kinase activity determined.

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DETAILED DESCRIPTION OF THE PREFERRED EMBODIMENTS

The present invention provides compounds of general Formula (I):



5

wherein

10 R_1 is a saturated or unsaturated hydrocarbon chain of from about 9 to about 26 carbon atoms and which is optionally carries one or more of a oxa, thia, hydroxy, hydroperoxy, epoxy and peroxy substitution;

each of R_2 , R_4 and R_6 is selected from O_2 , NO , NO_2 , $S(O)_x$, $C(H)_y$, H , $COOH$, $P(X)_\delta(Y)$,

15 $N(H)_z$, $C=O$, OH , $\text{---}\overset{\overset{O}{\parallel}}{C}\text{---}NH\text{---}$, C_{1-6} alkyl, C_{1-6} alkoxy, amino, mono-acid di- C_{1-6} alkylamino, C_{1-6} alkylthio, $S(O)_x\text{---}C_{1-3}$ alkyl, C_{1-6} alkoxycarbonyl, halo selected from fluoro, chloro, bromo and iodo, oxo, amidino and guanidino, C_{2-12} alkenyl, C_{2-12} alkynyl, aryl, heteroaryl and cyano, wherein x and z are 0, 1 or 2 and y is 0, 1, 2 or 3 and X is O , S or NR_8 , Y is OR_9 , SR_{10} or $NR_{11}R_{12}$ and R_8 , R_9 , R_{10} , R_{11} and R_{12} are selected from H , alkyl, alkenyl, alkynyl, aryl and heteroaryl, δ is 0 or 1;

20

each of R_3 , R_5 and R_7 is respectively $[(CH_2)_j (COOH)_k]_l$, $[(CH_2)_m (COOH)_n]_o$ and $[(CH_2)_p (COOH)_q]_r$, wherein each of j , m and p is 0, 1, 2, 3, 4, 5 or 6, each of k , n and q is 0, 1 or 2, and each of l , o and r is 0 or 1,

25 each of c , i and f is 0 or 1 or 2; and

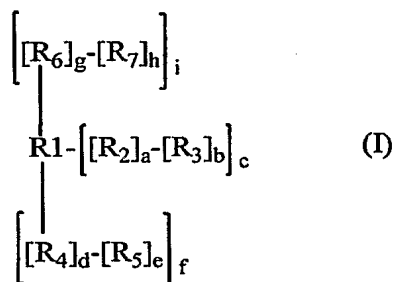
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each of a, d and g is 0 or 1 or 2;

each of b, e and h is 0 or 1 or 2.

5

More particularly, the present invention contemplates a method for the treatment or prophylaxis of a condition selected from a NFκB related or associated condition, a PKCβ related or associated condition, vascular or immunological conditions such as diabetes, inflammation, neurological conditions, cardiovascular disease and pain in a subject said
10 method comprising administering to said subject an effective amount of a compound having the structure of Formula (I):



15 wherein

R₁ is a saturated or unsaturated hydrocarbon chain of from about 9 to about 26 carbon atoms and which is optionally carries one or more of a oxa, thia, hydroxy, hydroperoxy, epoxy and peroxy substitution;

20

each of R₂, R₄ and R₆ is selected from O₂, NO, NO₂, S(O)_x, C(H)_y, H, COOH, P(X)_δ(Y),

N(H)_z, C=O, OH, $\text{---}\overset{\text{O}}{\parallel}\text{C}\text{---}\text{NH}\text{---}$, C₁₋₆ alkyl, C₁₋₆ alkoxy, amino, mono-acid di-C₁₋₆ alkylamino, C₁₋₆ alkylthio, S(O)_x-C₁₋₃ alkyl, C₁₋₆ alkoxy-carbonyl, halo selected from fluoro, chloro, bromo and iodo, oxo, amidino and guanidino, C₂₋₁₂ alkenyl, C₂₋₁₂ alkynyl,

25 aryl, heteroaryl and cyano, wherein x and z are 0, 1 or 2 and y is 0, 1, 2 or 3 and X is O, S

- 15 -

or NR₈, Y is OR₉, SR₁₀ or NR₁₁R₁₂ and R₈, R₉, R₁₀, R₁₁ and R₁₂ are selected from H, alkyl, alkenyl, alkynyl, aryl and heteroaryl, δ is 0 or 1;

each of R₃, R₅ and R₇ is respectively [(CH₂)_j (COOH)_k]_l, [(CH₂)_m (COOH)_n]_o and [(CH₂)_p (COOH)_q]_r, wherein each of j, m and p is 0, 1, 2, 3, 4, 5 or 6, each of k, n and q is 0, 1 or 2, and each of l, o and r is 0 or 1,

each of c, i and f is 0 or 1 or 2;

each of a, d and g is 0 or 1 or 2;

each of b, e and h is 0 or 1 or 2;

said administration being for a time and under conditions sufficient to prevent the condition or to ameliorate one or more symptoms of the condition.

The compound of Formula (I) may comprise when i, c and f are 0, a straight hydrocarbon chain such as that shown in Formula (II):



which represents a hydrocarbon chain of a'' carbons in length from about 9 to about 26 which hydrocarbon chain is saturated or unsaturated and which carries one or more of a oxa, thia, hydroxy, hydroperoxy, epoxy and/or peroxy substitution;

a' may be 0, 1, 2 or 3.

In one preferred embodiment, two of i, c or f is 0 and one of the remaining i, c or f is 1 resulting in the compound of Formula (III) using the example of i and f being 0:



- 16 -

wherein R_1 , R_2 , R_3 , a and b are as defined above.

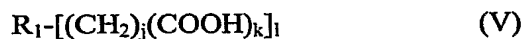
In another preferred embodiment, a in Formula (III) is 0 and b is 1 resulting in compound
5 of Formula (IV):



wherein R_1 and R_3 are as defined above.

10

Given that R_3 is $[(CH_2)_j(COOH)_k]_l$, Formula (IV) can be represented as a compound of
Formula (V):



15

wherein R_1 , j , k and l are as represented above.

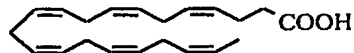
In a preferred embodiment, l is a saturated or unsaturated fatty acid. In another preferred
embodiment the saturated or unsaturated fatty acid carries one or more of a β -oxa, α -oxa,
20 γ -oxa, β -thia, α -thia, γ -thia, hydroxy, hydroperoxy, epoxy, peroxy, peracetyl or other
protected hydroperoxy substitution. Substitutions may be at the level of a carbon atom or
hydrogen atom.

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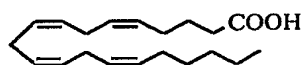
Examples of compounds of Formula (V) include:



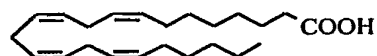
18:3n-3



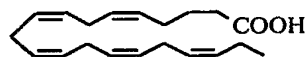
22:6n-3



20:4n-6

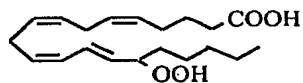


23:4n-6



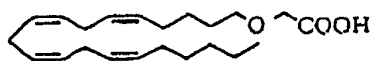
20:5n-3

5 Examples of when R_1 comprises a substitution include:

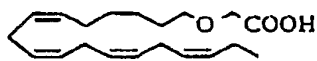


15-OOH-20:4n-6

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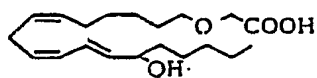
β -oxa-23:4n-6 (MP3)



β -oxa-21:4n-3 (MP7)



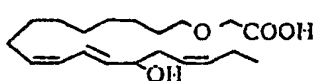
β -oxa-21:3n-6 (MP4)



16-OH- β -oxa-21:3n-6 (TR1)



β -oxa-21:3n-3 (MP5)



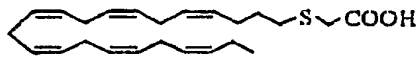
16-OH- β -oxa-21:3n-3 (TR2)



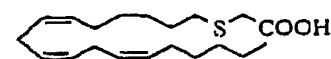
β -oxa-25:6n-3 (MP6)



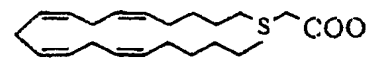
β -thia-21:0 (MP2)



β -thia-25:6n-3 (MP14)



β -thia-21:3n-6 (MP9)

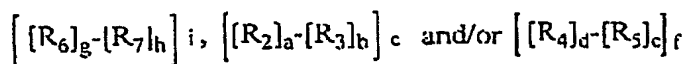


β -thia-23:4n-6 (MP8)



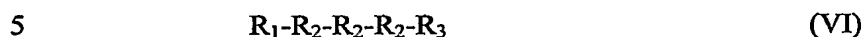
β -thia-21:3n-3 (MP10)

5

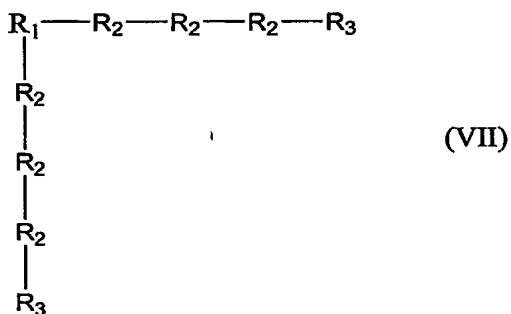


- 19 -

are presented in multiple forms then the multiple forms may be represented linearly. For example, if l and f are each 0, a is 3, b is 1 and C is 1, then the compound may be represented as in Formula (VI):



If, on the other hand, c is 2, then the compound is represented as Formula (VII):



10

In one non-limiting example, in the case when the compound is a carboxymethyl derivative, then the values in Formula (I) are as follows:

i is 0, each of c and f is 1, each of a and d is 0 and each of R_3 and R_5 is $[(CH_2)_j (COOH)_k]_l$ and $[(CH_2)_m (COOH)_n]_o$, respectively where, in one example,

each of j and m is 0,

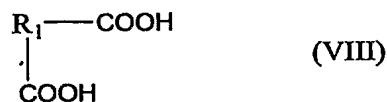
each of l and o is 1; and

20

each of k and n is 1,

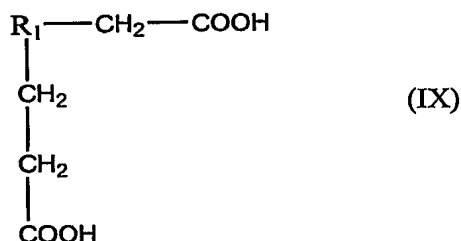
resulting in a compound of Formula (VIII):

- 20 -



More commonly, however, j may be 1, and m may be 2 resulting a compound of Formula (IX):

5



Reference to "from about 9 to about 26 carbon atoms" includes 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22, 23, 24, 25 and 26 carbon atoms.

10

The compound of Formula (I) may have each of i, c and f as 0 (zero), two of i, c and f as 0 (zero) or one of i, c and f as 0 (zero); or each of i, c and f as 1; two of i, c and f as 1 or one of i, c and f as 1; or each of i, c and f as two, two of i, c and f as two, or one of i, c and f as two;

15

The compound of Formula (I) may have each of g, a and d as 0 (zero), two of g, a and d as 0 (zero) or one of g, a and d as 0 (zero); or each of g, a and d as 1; two of g, a and d as 1 or one of g, a and d as 1; or each of g, a and d as two, two of g, a and d as two, or one of G, a and d as two;

20

The compound of Formula (I) may have each of h, b and e as 0 (zero), two of h, b and e as 0 (zero) or one of h, b and e as 0 (zero); or each of h, b and e as 1; two of h, b and e as 1 or one of h, b and e as 1; or each of h, b and e as two, two of h, b and e as two, or one of h, b and e as two.

25

Those aspects of the present invention cover naturally occurring PUFAs as well as

synthetic, modified or derivitized PUFAs. Furthermore, modified PUFAs encompassed by Formulae (I) through (VIII) include naturally occurring or synthetic, derivitized PUFAs conjugated to an L- or D-amino acid or amino acid analog or a sequence of amino acids such as a protein. The latter aspect includes proteins in the form of cytokines, growth
5 factors, proteases, enzymes, apoptotic proteins and pro-survival proteins.

Examples of L-amino acids include alanine, arginine, asparagine, aspartic acid, cysteine, glutamine, glutamic acid, glycine, histidine, isoleucine, leucine, lysine, methionine, phenylalanine, proline, serine, threonine, tryptophan, tyrosine and valine.

10

Examples of chemical analogs of amino acids include but are not limited to α -aminobutyric acid, α -amino- α -methylbutyrate, aminocyclopropane-, carboxylate, aminoisobutyric acid, aminonorbornyl-, carboxylate, cyclohexylalanine, cyclopentylalanine, D-alanine, D-arginine, D-aspartic acid, methylmethionine, D-cysteine,
15 N-methylnorleucine, D-glutamine, D-glutamic acid, methylornithine, D-histidine, N-methylphenylalanine, D-isoleucine, D-leucine, D-lysine, D-methionine, D-ornithine, D-phenylalanine, D-proline, D-serine, D-threonine, D-tryptophan, D-tyrosine, D-valine, D- α -methylalanine, D- α -methylarginine, D- α -methy lasparagine, D- α -methy laspartate, D- α -methylcysteine, D- α -methylglutamine, D- α -methylhistidine, D- α -methylisoleucine, D- α -
20 methylleucine, D- α -methyllysine, D- α -methylmethionine, D- α -methylornithine, D- α -methylphenylalanine, D- α -methylproline, D- α -methylserine, D- α -methylthreonine, D- α -methyltryptophan, D- α -methyltyrosine, D- α -methylvaline, D-N-methylalanine, D-N-methylarginine, D-N-methy lasparagine, D-N-methy laspartate, D-N-methylcysteine, D-N-methylglutamine, D-N-methylglutamate, D-N-methylhistidine, D-N-methylisoleucine, D-
25 N-methylleucine, D-N-methyllysine, N-methylcyclohexylalanine, D-N-methylornithine, N-methylglycine, N-methylaminoisobutyrate, N-(1-methylpropyl)glycine, N-(2-methylpropyl)glycine, D-N-methyltryptophan, D-N-methyltyrosine, D-N-methylvaline, γ -aminobutyric acid, L-t-butylglycine, L-ethylglycine, L-homophenylalanine, L- α -methylarginine, L- α -methy laspartate, L- α -methylcysteine, L- α -methylglutamine, L- α -
30 methylhistidine, L- α -methylisoleucine, L- α -methylleucine, L- α -methylmethionine, L- α -

methylnorvaline, L- α -methylphenylalanine, L- α -methylserine, L- α -methyltryptophan, L- α -methylvaline, N-(N-(2,2-diphenylethyl)carbonylmethyl)glycine, and 1-carboxy-1-(2,2-diphenyl-ethylamino)cyclopropane.

- 5 Examples of cytokines include but are not limited to BDNF, CNTF, EGF, EPO, FGF1, FGF2, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGF10, FGF11, FGF12, FGF12, FGF13, FGF14, FGF15, FGF16, FGF17, FGF18, FGF19, FGF20, FGF21, FGF22, FGF23, G-CSF, GM-CSF, IFN α , IFN β , IFN γ , IL1, IL2, IL3, IL4, IL5, IL6, IL7, IL8, IL9, IL10, IL11, IL12, IL13, IL14, IL15, LIF, MCP1, MCP2, MCP3, MCP4, MCP5, M-CSF, MIP1,
10 MIP2, NGF, NT 3, NT4, NT5, NT6, NT7, OSM, PBP, PBSF, PDGF, PF4, RANTES, SCF, TGF α , TGF β , TNF α , TNF β , TPO, VEGF, GH, insulin and the like.

- Examples of apoptotic proteins include but are not limited to A1, A9, A20, A46R, A52R, A53, A238L, Aac11, AATF, AATYK, ABIN1, ABIN-1, ABIN2, Acidic Sphingomyelinase,
15 Acinus, Act1, ACT2, Activin, AD3LP, AD5, ADAR, adrenomedullin, aggrecan, AMAM17, 33, A11, AIF, AILIM, AIM2, AIR, AITR, Akt, ALCAM, ALG2, ALG3, ALG4, ALP, Alix, Armadillo, AMAC1, AMH, AMID, Amida, angiotensinogen, Ankyrin, ANT1, AO7, AP1, Apaf-1, APC, APC2, APCL, APE1820, APJ, APO-1, APO-2, APO-3, Apopain, APP1, APP2, Apr, APRIL, ARA54, ARC, ARF, arkadia, ARIH1, 2, ASC, Ash2,
20 Ask1, Ask2, ASPP1, ASPP2, AT2R1, AT2R2, ATAR, ATF1, ATF2, ATF3, ATF4, ATM, atona, ATR1, AUF1, Aven, AVP, AvrA, AvrBsT, Axi, Axin, Axin 2, Axi, b-catenin, b-TrCP, B28R, B7-1, B7-2, B7h2, B7RP1, Bach2, Bad, BAFF, BAG -1, -2, -3, -4, -5, Bak, BALF1, Bam32, BAP-1, BAP31, BAP29, BAR, BARD1, BAT3, Bax, BBc3, BCA1, BCAN, Bcl-2, BCL2, Bcl-3, Bcl-10, BCL10, Bcl-G, Bcl-Rambo, Bcl-w, Bcl-x, beclin,
25 BEHAB, BERP, Bfl-1, BFL1, BG1, BG2, BG4, BG5, BHP1, BHRF1, BI-1, Bid, Bif-1, Bik, Bis, Bim, Bimp-1, Bimp1, Bimp2, Bimp3, BIR1, BIRP, BL-CAM, BLC, Blk, BLNK, BLR1, BLyS, BMI-1, BmP109, BNIP3, BNIP3a, BNIP3L, Bok, bone sialoprotein, bonus, Boo, BPI, BRAL1, BRAG-1, BRAP, Bravo, BRCA1, BRN3a, BRN3b, BRN3c, brevican, BPR, BSAC, BUFFY, C1q, C1r, C1s, C2, C3, C4a, C4b, C5, C6, C7, C8a, C8b, C8g, C9,
30 C1qBP, C3aR, C4BP_{a,b}, C5R1, CR2, CIITA, C5L, c-E10, c-FLIP, c-Fms, c-Fos, c-IAP1, cIAP1, c-IAP-1, c-IAP2, cIAP2, c-IAP-2, c-Jun, c-Myc, c-Rel, Cactus, CAD, cadherin, E,

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N, P, VE, calcineurin, CARD4,, CARD7, CARD9, CARD10, CARD11, CARD12,
 CARD14, CARDIAK, Carma1, CARMA-1, CARMA2, CARMA3, CARMA, CARMEN,
 CAP1, CAR1, CART1, CAS, CAS-L, Caspase -1, -2, -3, 4, -5, -6, -7, -8, -9, -11, -12, -13, -
 14, Casper -1, -2, -3, -4, -5, -6, -7, -8, -9, -10, -11, -12, -13, -14, -15, -16, -17, -18, -19, -20,
 5 -21, -22, -23, -24, -25, -26, -27, -28, CASH, CBL, CBL-B, CBL-C, CC-CKR-6, CCF,
 CCL, CCPI, CCRs, CD2, CD3, CD4, CD5, CD6, CD7, CD8, CD9, CD11, CD14, CD18,
 CD19, CD20, CD21 (CR2), CD22, CD23, CD25, CD27, CD27L, CD28, CD28LG1,
 CD28LG2, CD29, CD30, CD31, CD32, CD33, CD34, CD35, CD36, CD40, CD40L,
 CD41, CD43, CD44, CD45, CD46, CD47, CD48, CD49, CD50, CD53, CD54, CD55,
 10 CD56, CD58, CD59, CD61, CD62E, L, H, CD66, CD63, CD64, CD66a - e, CD67, CD70,
 CD72, CD74, CD79a, b, CD80, CD84, CD85a -m, CD86, CD88, CD89, CD90, CD92,
 CD94, CD95, CD96, CD97, CD99, CD100, CD101, CD102, CD104, CD105, CD106,
 CD108, CD112, CD115, CD116, CD117, CD119, CD120a, b, CD121a, b, CD122, CD123,
 CD124, CD125, CD126, CD127, CD128a, b, CD130, CD131, CD132, CD134, CD135,
 15 CD136, CD137, CD140a, CD140b, CD143, CD144, CD146, CD147, CD148,
 CD150, CD151, CD152, CD153, CD154, CD155, CD158a-z, CD159, CD160, CD161,
 CD162, CD166, CD178, CD180, CD183, CD184, CD195, CD197, CD207, CD229,
 CD244, CDC2, CDC25, CDC42, CDK1, CDK2, CDK5, CDM, CEA, CEAL, CEACAM1,
 6, C/EBP, CED1, CED2, CED3, CED4, CED5, CED6, CED7, CED8, CED9, Ced-9,
 20 CED10, CED11, CED12, CED, CEP-1, CES1, CES2, CES3, CETP, CeTRAF, Cezanne,
 CGR19, CGRP, Che1, Che-1, CHFR, chemokines, CHOP, CHUK, cIAP1, cIAP2, c-IAP1,
 c-IAP2, c-IAP-1, c-IAP-2, CIDE -A, -B, CIKS, CIN85, CIP-1, CIPER, CISK, Ckb-8,
 CKR1, 2, 3, 4, 5, CKRL1, Clan, CLAP, CLARP, CMD1, CMH1, CMKBR1, 2, 3,, 4, 5, 6,
 CMPD1, conductin, Cop9 subunit 3, COP11, COPS3, COPS5, COT, COX-1, COX-2,
 25 CPAN, CPP32, CPZ, CRADD, CRAF1, CR8, CREB, CREM, Crk-II, crinkled, crmA,
 crmB, CSBP1, CSMF, CSN3, Csp -1, -2, -3, CSPG2, 3, Csx, CTACK, CTAP3, CTGF,
 CTLA4, cytochrome c, cytosolic PL A2, CXCLs, CXC-R3, DAAM1, Dad1, DAD-1,
 Damm, DAP1, DAP3, DAP5, DAP12, DAP kinase 1, DAPP1, DAYDREAM, DAXX,
 Dborg1, dCAD, DCCCK1, DCP1, Dcp-1, DCP2, Dcp-2, DcR 1,2,3, DD2, Decay, DED,
 30 DEDAF, DEDD, DEDD2, dedpro1, defensin, DEFT, dFADD, DFF , DFF35, DFF40,
 DFF45, DG17, Diablo, DIAP1, DIAP2, Dickkopf, DIF, DIF2, DIHA, DIK, Drosophila

- 24 -

IKK, PKCdelta-interacting protein kinase, DIO1, DIP, disshevelled, diubiquitin,
 DKK1,2,3,4, DLAK, DLK, DMDL, DNase II, Diva, DONG1, Dorsal, DP1, DP2, DP5,
 Drob1, DRP-1, DocA, dock188, Dok1, Doom, dorfin, DR3,4,5,6, DRAK 1, 2, DREAM,
 DREP -1, -2, -3,-4, DrICE, DRONC, DRP1, DTR, DTS, DUSP, E1.1, E1B 19K, E10,
 5 E2Fs, E4BP4, E4ORF4, E8, E4, E48, E3RS, eae7, Ear7, EBAF, EBI1, EBP1, EBI6,
 ECSIT, EDA, EDAR, Edradd, EFP, EGL1, Egr1, 2, 3, EHF, eIF-2aK, Eiger, ELAM,
 ELF2, ELK1 -4, EMR1, ENA78, Endofin, Endoglin, Endophilin B1, endothelin, ENG,
 eNOS, eotaxin 1,2, ERN1, ERICE, ES18, Ets-1, -2, ER81, ErbAa, ERG, ERM, ESE2,
 Eskine, ETV1, 2,3,4,5,6, exodus-1, 2, 3, FADD, Fas associated via death domain, FAF1,
 10 FAIM, FAN, FANCC, Fas, FAST, FAT10, fb1, FCAR, FELL, FEM-1, FEM-2, FHR1, 2,
 3, 4, 5, FKBP, FIGF, FIL1d, e, eta, zeta, FIP2, FIP-2, FIP3, FIP-3, FKSG2, FIST,
 FKHL12, FKHR, FKHL1, FLAME-1, FLAME-3, FLAME3, FLASH, FLDED-1, FLI-1,
 FLI1, FLICE, FLICE2, FLICE-2, FLIP, FLT3L, Fliz1, Fln29, Fms, Fnk, fortilin, Fos,
 FOXO1A, FOXO3A, FOXE3, FPV039, Fra1, Fra2, Fractalkine, FRAP, FREAC8,
 15 Frizzled, Fzd, Fz, FRING, FRP1, 2, 3, FRP1(ATR), frpHE, FRZB-PEN, Fsp27, FUS,
 FUS6, Fusin, FXY, FY, G-coupled receptors, G10P1, G25K, G4R, G6C, G6E, GADD34,
 GADD45, GADD153, GATA1,2,3,4,5,6, GBP2, GCP2, GDFs, geisolin, Gfi-1, Gfi1,
 GFRP1, GILZ, gingipain, GTR, GL50, glycodelin A, GM2A, gp34, GPR5, GPR9, GPR-
 9-6, Granzyme B, Grim, GRMP, Groa, Grob, GRS, GSKbeta, H2TF1, H731-like, Hakai,
 20 HB-EGF, Hck, HF1, HFB30, HFL3, HHARI, hIAP-1, hIAP1, Hid, HIF1 alpha, HIP1,
 HIP116, HIPPI, HIPK1,2,3, histamine receptors, HIVEP1, 3, HIV-EP1, HLTF, HM85,
 HM89, HM145, HMR, HNRPD, HRD1, Hrk, HtrA2, Huntingtin, HVEM, HVEML, HYP,
 IAP-1, IAP1, IAP2, IAP, iAPP, ICAD, ICBP90, ICE, ICEBERG, ICE-LAP3, ICE-LAP6,
 ICErel-II, ICErel-III, Ich1, ICH-1, Ich2, ICH-2, Ich3, ICH-3, ICOS, I-TRAF, I-FLICE,
 25 IEX-1m IFI, IFIT1, 2, 3, 4, IFP35, IgE Fc Receptor, IGF1 and its receptor, IGFBP-3,
 IKAP, Ikaros, IKB-1, Ikb a, b, e, IKKAP1, IKK 1,2, IKK a,b, IKKg, interleukins,
 interleukin receptors, IL1 antagonist, anti-IL1, IL1RacP, IL8R1, ILA, ILC, ILP, ILP-1,
 ILP-2, ILT1-11, ING1, ING2, ING3, Inhibin, INK4, INK4A, integrin, IP10, INP10, IP30,
 Ipaf, IRAK, IRAK2, IRAM-M, IRE1, RE1p, IRE, IRF, IRTA1-5, ISGF3g, ITA, It, Jab1,
 30 Jak1, 2, 3, JDP2, JIK, JN, K, K13, KARAP, KBF 1, 2, 3, KDS, KE05, KET, kf-1, KIAP,
 Killer, KIR2DL1-5, KIR2DS1-6, KROX2, L-Myc, lactalbumin alpha, LAG1, LAIR1,

LALBA, LAM, LAP1, LAP3, LAR, LARD, LARC, LATS1, 2, LBP, Lck, LCP2, LD78b,
 LEFTY, LESTR, Leu1, Leu8, Leu14, leukotactin, LFA3, LFG, LICE, LICE2, LIF,
 LIGHT, LIR1, 2, 3, 4, 5, 6, 7, 8, Livin, LMP1, LMW5-HL, LOK, Lot1, LRDD, LRP, Low
 affinity NGFR, LTa, LTb, LTbR, LTP2, Ly63, lymphotactin, Ly1, Lyf1, Lysozyme, Lyt-
 5 10, LYVE1, LZK, M11, M159L, M160L, MA-3, MACH, Mad, Mad3, MADD, Maf, c-
 Maf, makorin, MAL, MALT, MAP-1, MAPKKKKs, MAPKKKs, MAPKKs, MAPKs,
 Math1, Max, MBD4, MBLR, MBP1, MCL1, Mch2, Mch3, Mch4, Mch5, Mch6, MCP1, 2,
 3, MCP-1, Mda-7, MD-1, MD-2, Mdm2, Mdm4, MdmX, MDP62, mE10, MEF2a,
 MEKKs, Mel-18, MEMD, Meprin, metacaspase, MIC1, MID1, MIF, MIG, MIHC, MIP1,
 10 2, 2a, 2b, MIP-T3, MIR, MIS, MITF, MKK6, MKL1, MKP1, ML-1, ML-IAP, MLN64,
 MLX, MMP-1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16, MNDA, MNT, Mob1, mod (mdg4),
 MORT1, MPIF1, 2, MRFP, MRIT, Msx1, Msx2, MTAP44, Mtd, mTOR, MUC1, MUC2,
 MUL, MURF-1,2,3, myp-nop30, MxA, MxB, Mxi1, Mxi2, MYAK, Myc, MyD88,
 MyD118, MYLK, myoblast city, N-Myc, NAF1, NAIP, NALP1, NALP2, NAP2, NBAK3,
 15 Nbk, NBS1, NCA, NCAM, NCC1, 2, 3, 4, NDG1, Neural Sphingomyelinase, Neuralin,
 NEMO, neogenin, Neurotactin, neurocan, NF-kB, NF-X1, NFATs, NFIL3, NFIL6,
 NFkB1, 2, NIP1, NIP2, NIP3, NIPK, NIK, Nix, NKAT1-9, NKX2-5, nNOS, Notch,
 NOD-1, NOD-2, nop30, Nor-1, NOS2, NOS2B, NOS3, Nov, Noxa, NP10, Np95, Npc2,
 NPY3R, Nr-CAM, NR3, NR13, Nr-13, NRAGE, NRIF1, nucleolin, Nur77, NY-REN-64,
 20 OCIF, ODF, ODFR, OIAS, ORF16, osteoprotegerin, OSX, OX40, OX40L, OPG, OPGL,
 Osi, osteonectin, osteoponti, p14, p16, p33ING1, p35, p38, p49, p49, p55, p52, p53,
 p53AIP1, p53DINP1, p55, p60, p62, p62Dok, p63, p65, p73, p75NTR, p84, p100, p105,
 p193, p202, PAC1, PACAP, PACT, PAF400, PAG-3, PAG608, PAK1, PAK2, PAK3,
 PAP1, PAR4, paracaspase, PARC, Park2, parkin, PARP, PAX2, 3, 5, 8, PBEF, PBP, PD1,
 25 PDGF, PEA15, Pellino, PERK, PERP, PEK, Pelle, PEX10, PF4, PGRP, PI3K, Pidd, PIK-
 1, PLAB, Plk, Plk3, PKC, PKR, PKY, PLAGL1, PLAIDD, PLA2, PLC, PLD, Pli, Pml,
 PMP41, POSH, PP1A, PP14, PP2Ca, PRKR, PRSS25, polycystin 1, porimin, PRG1, Prk,
 PRL, prolactin receptor, PS -1, -2, PSCA, PSMD11, 12, 13, PSP-C, PSK, PSSALRE,
 PTEN, PTK1, PTPs, PTP1C, PTP2C, PTP1G, PTPL1, PU.1, puckered, Pum, Q2/2, Rac,
 30 RAI, RANTES, RAX, Rb, Relish, RELT, Raf, RANK, RANKL, RAIDD, RBBP6, RBQ1,
 Rcm, Reaper, RelA, relaxin H1,H2,H3, RelB, Requiem, RFP, RFPL-1,2,3, RGS, RhoA,

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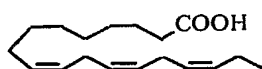
RICK, RIG-G, Ro52, Ro 60kDa, ROC-1, 2, RORgamma, ROX, RIFF, RIP, RIP2, RIP3,
 RNM561, RNF, RP-8, RP8, RP105, Rpr, RRP5, RYBP, S9, S152, SAG, Salvador, SAP1,
 SAPK2A, Sara, SARP 1,2,3, Sav, Sca2, SCA-2, SCC-S2, SCF, SCDGF, SCM1, 1a,
 Scythe, SDF1, selectin L, E, P, SENP1, SENP2, sentrin/SUMO-specific protease, SETA,
 5 SFRP1, 2, 3, 4, 5, SFTP2, SFTPC, SGK, SGL, SGN5, SH2D1A, SHP1, 2, Siah, SIMPL,
 SIP27, SIP18, SIR2, SIVA, SLC, SLK, SLP-65, SLP-76, SLUG, Smac, SMADs,
 SMARCA3, SMN, SMT 3A, B, 3C, SNAIL, SNF2L3, SODD, somatostatin, Son3, SOX9,
 SP5, SP-C, SPARC, Sphigomyelinase, Smase, SPOP, SPP1, SPRK, Spatzle, SFRP1,2,5,
 SS-56, SSA, SSA1, SSA2, ST2L, stabilin 1,2, STATs, STCP1, STG6, STEP, STM-2,
 10 Stra3, STRICA, Substance P, SUMO1, survivin, SYK, SY, T cell receptor, T2BP, T6BP,
 TAB1, Tab2, Tabby, TACI, TACTILE, Tag7, tachykinin, TAJ, TAK1, Tak1, TALL-1,
 TANK, TAO1, TAO2, TARC, TBX1,2,3,4,10,18,19,20,21,22, TCA3, TCA-3, TC1, TC2,
 TCR, TCTP, TDAG51, TEAP, TECK, TEGT, TEL, (TEL1), TEL2 (TELb), telokin,
 TERF, TFT, TGb, TGFbeta 1,2,3, THG1, THRa, Thy-1, TIA1, TIAP, TIEG, TIF1,
 15 TIFgamma, TIL6, TIMP1,2,3, TIP49, Tip60, TIRAP, TIS, TLRs, TLS, TMS1, TNFa,
 TNFAIP3, A20, TNFAIP6, TNFb, TNF-C, TNFR1, TNFR2, TNFR-II, TNFRSF1-19, Toll,
 Tollo, Tollip, TONEBP, Toso, Tp44, TPL-2, TR3, TR2L, TRABID, TRADD, TRADE,
 TRAF1, TRAF1(Dm), TRAF2, TRAF2(Dm), TRAF3, TRAF4, TRAF5, TRAF6,
 TRAF6(Dm), TRAFamn, TRAIL, TRAIL-R2, TRAMP, TRANCE, TRC8, TRIAD1, 3,
 20 TRIF, TRIM, TRIP15, TRF-1, TRF-2, TRF1, TRF2, traube, TRDL-1, TRG, TRH,
 TRICK2, TRIP, Tristetraproline, TROY, TRRAP, TSC-22, TSC-22R, TTRAP, Tube,
 TUCAN, TWEAK, TX, TXBP151, TY, Tyk, UBCH7BP, UL36, UL37, Ulp, Unc5,
 UNC5h3, Urinary, stone protein (SPP1), USP7, usurpin, uterophi, vasopressin, Vav, vav1,
 vav2, vav3, vav-1, vav-2, vav-3, versican, vICA, VIAF1, vBcl-2, VEGI, VEGF,
 25 Ventroptin, VG-1, VG71, VHR, v-IAPs, VI, Warts, Wengen, WIG1, WISP-1, 2, 3, Wnt,
 WSL-1, WT1, WW45, WWOX, XAF1, XAP4, XCL1, 2, XEDAR, XIAP1, xRI, xRII,
 XICE, XICEa, XICE, Yama, YopJ, YY1AF, Zac, Zac1, ZAP70, ZBP89, zf3, ZFP26,
 ZFP127, ZH-DR, ZNF40, 124, 148, as TFs, ZNF144, 147, 179, 313, 364 as RING, ZIP-
 kinase, ZPR, 18 wheeler, 24.6K Glu/Pro-rich, 4-1BB, 4-1BBL, 4-1BB ligand, 53BP2,
 30 7TM.

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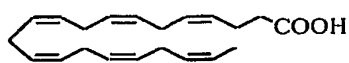
Examples of pro-survival proteins include but are not limited to, Bcl-2, Bcl-XL, Mcl-1 and A1.

Examples of PUFAs contemplated by the present invention include

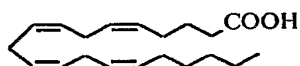
5



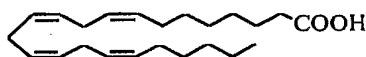
18:3n-3



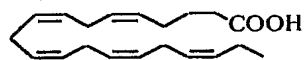
22:6n-3



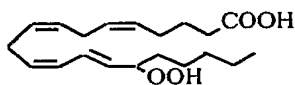
20:4n-6



23:4n-6

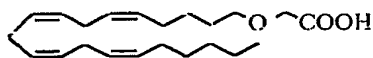


20:5n-3



15-OOH-20:4n-6

Natural PUFA and hydroperoxy derivative



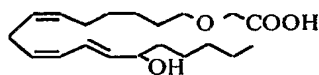
β -oxa-23:4n-6 (MP3)



β -oxa-21:4n-3 (MP7)



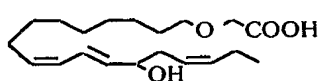
β -oxa-21:3n-6 (MP4)



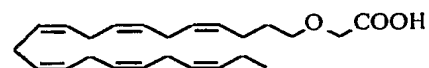
16-OH- β -oxa-21:3n-6 (TR1)



β -oxa-21:3n-3 (MP5)



16-OH- β -oxa-21:3n-3 (TR2)



β -oxa-25:6n-3 (MP6)

MP series, β -oxa compounds

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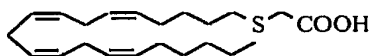
β -thia-21:0 (MP2)



β -thia-25:6n-3 (MP14)



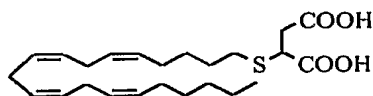
β -thia-21:3n-6 (MP9)



β -thia-23:4n-6 (MP8)

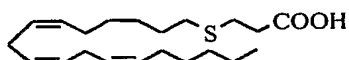


β -thia-21:3n-3 (MP10)

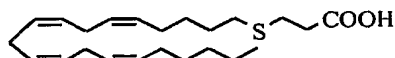


α -carboxymethyl- β -thia-23:4n-6 (MP15)

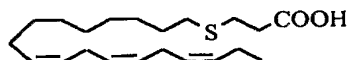
MP series, β -thia compounds



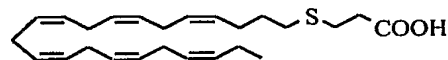
γ -thia-22:3 (n-6)



γ -thia-24:4 (n-6)

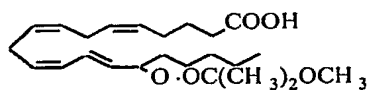


γ -thia-22:3 (n-3)

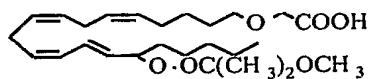


γ -thia-25:6 (n-3)

MP series, γ -thia compounds



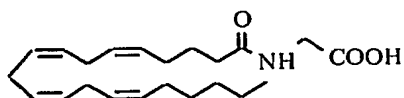
15-OOC(CH₃)₂OCH₃-20:4n-6 (MP16)



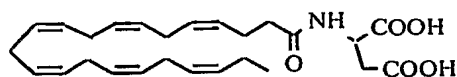
15-OOC(CH₃)₂OCH₃- β -oxa 23:4n-6 (MP17)

MP series, protected hydroperoxy compounds

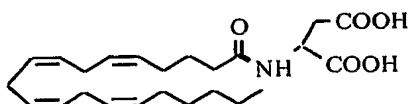
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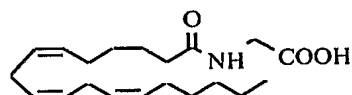
20:4n-6 Gly (PT1)



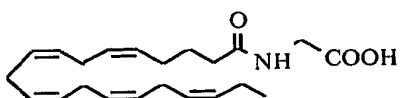
22:6n-3 Asp (PT6)



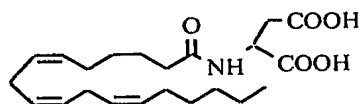
20:4n-6 Asp (PT2)



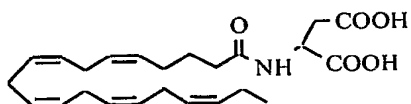
18:3n-6 Gly (PT7)



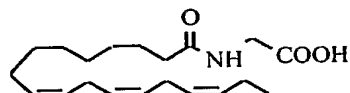
20:5n-3 Gly (PT3)



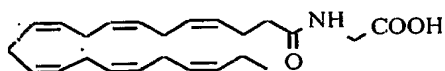
18:3n-6 Asp (PT8)



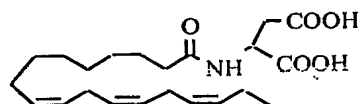
20:5n-3 Asp (PT4)



18:3n-3 Gly (PT9)



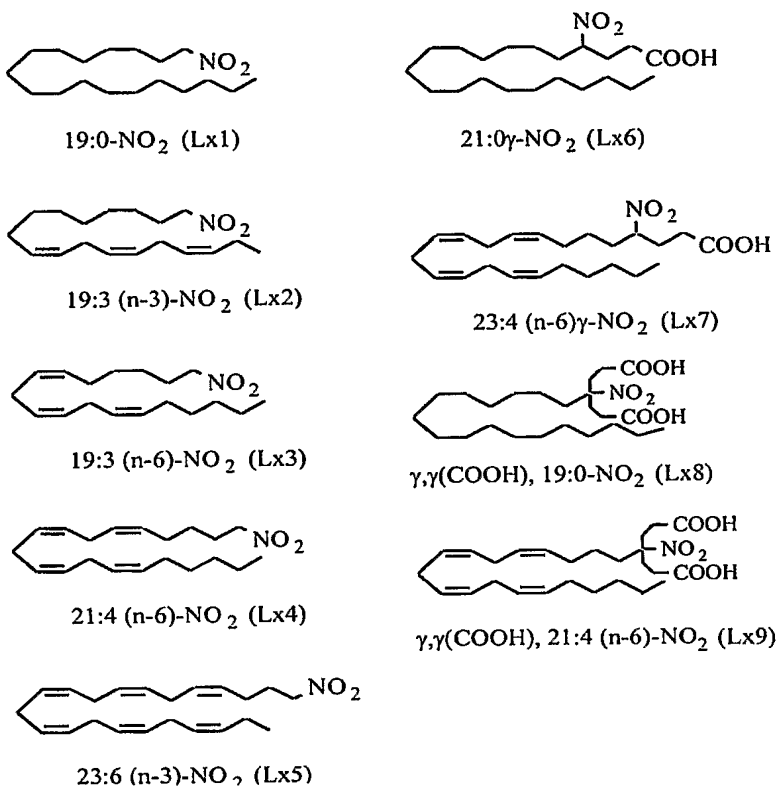
22:6n-3 Gly (PT5)



18:3n-3 Asp (PT10)

PT series: PUFA -amino acid conjugates

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LX series, nitroanalogues of fatty acids

The present invention is directed *inter alia* to the treatment of *inter alia* pain, cancers, PKC and/or NFκB-associated or related conditions, vascular and/or immunological conditions, inflammatory conditions, neurological conditions and infection.

Other compounds contemplated by the present invention include β-oxa 23:0, β-thia 23:0, β-oxa 23:4 (n-6), β-oxa 21:3 (n-6); β-oxa 21:3 (n-3), β-oxa 25:6 (n-3), β-oxa 21:4 (n-3), β-thia 23:4 (n-6), β-thia 21:3 (n-6), β-thia 21:3 (n-3), γ-thia 24:4 (n-6), γ-thia 22:3 (n-6), γ-thia 22:3 (n-3), β-thia 25:6 (n-3), α-CH₂CO₂H-β-thia 23:4 (n-6), 15-OOCMe₂OMe 20:4 (n-6), 15-OOCMe₂OMe β-oxa 23:4 (n-6), 13-OH-β-oxa 21:3 (n-6), 13-OH-β-oxa 21:3 (n-3), 20:4 (n-6)-gly, 20:4 (n-6)-asp, 20:5 (n-3)-gly, 20:5 (n-3)-asp, 22:6 (n-3)-gly, 22:6 (n-3)-asp, 18:3 (n-6)-gly, 18:3 (n-6)-asp, 18:3 (n-3)-gly, 18:3 (n-3)-asp, 19:0-NO₂, 19:3

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(n-3)-NO₂, 19:3 (n-6)-NO₂, 21:4 (n-6)-NO₂, 23:6 (n-3)-NO₂, γ -NO₂ 21:0, γ -NO₂ 23:4 (n-6) and γ,γ (COOH), 21:4 (n-6)NO₂.

The present invention is particularly directed to the treatment of pain including *inter alia*
5 neuropathic or neurological pain, chronic pain, acute pain, migraine, headache
inflammatory pain, postoperative pain, pain due to multiple sclerosis, Parkinson's disease
or other nuerological or autoimmune disorder or following or during periods of anxiety,
delayed onset muscle soreness, burns or during or following infection or a convulsion,
post-poliomyelitic pain, bipolar disorder, panic attack or epilepsy.
10
Neurological disease states which can be treated in accordance with the present invention
include depression, including major depression (single episode, recurrent, melancholic),
atypical, dysthymia, subsyndromal, agitated, retarded, co-morbid with cancer, diabetes, or
post-myocardial infarction, involutional, bipolar disorder, psychotic depression,
15 endogenous and reactive, obsessive-compulsive disorder, or bulimia. In addition,
NAALADase inhibitors can be used to treat patients suffering from pain (given alone or in
combination with morphine, codeine, or dextropropoxyphene), obsessive-compulsive
personality disorder, post-traumatic stress disorder, hypertension, atherosclerosis, anxiety,
anorexia nervosa, panic, social phobia, stuttering, sleep disorders, chronic fatigue,
20 cognition deficit associated with Alzheimer's disease, alcohol abuse, appetite disorders,
weight loss, agoraphobia, improving memory, amnesia, smoking cessation, nicotine
withdrawal syndrome symptoms, disturbances of mood and/or appetite associated with
pre-menstrual syndrome, depressed mood and/or carbohydrate craving associated with pre-
menstrual syndrome, disturbances of mood, disturbances of appetite or disturbances which
25 contribute to recidivism associated with nicotine withdrawal, circadian rhythm disorder,
borderline personality disorder, hypochondriasis, pre-menstrual syndrome (PMS), late
luteal phase dysphoric disorder, pre-menstrual dysphoric disorder, trichotillomania,
symptoms following discontinuation of other antidepressants, aggressive/intermittent
explosive disorder, compulsive gambling, compulsive spending, compulsive sex,
30 psychoactive substance use disorder, sexual disorder, schizophrenia, premature ejaculation,

or psychiatric symptoms selected from stress, worry, anger, rejection sensitivity, and lack of mental or physical energy.

Other examples of pathologic, psychologic conditions which may be treated in accordance with this invention include, but are not limited to: Moderate Mental Retardation, Severe Mental Retardation, Profound Mental Retardation, Unspecified Mental Retardation, Autistic Disorder, Pervasive Development Disorder NOS, Attention-Deficit Hyperactivity Disorder, Conduct Disorder, Group Type, Conduct Disorder, Solitary Aggressive Type, Conduct Disorder, Undifferentiated Type, Tourette's Disorder, Chronic Motor or Vocal Tic Disorder, Transient Tic Disorder, Tic Disorder NOS, Primary Degenerative Dementia of the Alzheimer Type, Senile Onset, Uncomplicated, Primary Degenerative Dementia of the Alzheimer Type, Senile Onset, with Delirium, Primary Degenerative Dementia of the Alzheimer Type, Senile Onset, with Delusions, Primary Degenerative Dementia of the Alzheimer Type, Senile Onset, with Depression, Primary Degenerative Dementia of the Alzheimer Type, Presenile Onset, Uncomplicated, Primary Degenerative Dementia of the Alzheimer Type, Presenile Onset, with Delirium, Primary Degenerative Dementia of the Alzheimer Type, Presenile Onset, with Delusions, Primary Degenerative Dementia of the Alzheimer Type, Presenile Onset, with Depression, Multi-infarct dementia, Uncomplicated, Multi-infarct dementia, with Delirium, Multi-infarct Dementia, with Delusions, Multi-infarct Dementia, with Depression, Senile Dementia NOS, Presenile Dementia NOS, Alcohol Withdrawal Delirium, Alcohol Hallucinoses, Alcohol Dementia Associated with Alcoholism, Amphetamine or Similarly Acting Sympathomimetic Intoxication, Amphetamine or Similarly Acting Sympathomimetic Delusional Disorder, Cannabis Delusional Disorder, Cocaine Intoxication, Cocaine Delirium, Cocaine Delusional Disorder, Hallucinogen Hallucinoses (305.30), Hallucinogen Delusional Disorder, Hallucinogen Mood Disorder, Hallucinogen Posthallucinogen Perception Disorder, Phencyclidine (PCP) or Similarly Acting Arylcyclohexylamine Intoxication, Phencyclidine (PCP) or Similarly Acting Arylcyclohexylamine Delirium, Phencyclidine (PCP) or Similarly Acting Arylcyclohexylamine Delusional Disorder, Phencyclidine (PCP) or Similarly Acting Arylcyclohexylamine Mood Disorder, Phencyclidine (PCP) or Similarly Acting Arylcyclohexylamine Organic Mental Disorder NOS, Other or

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unspecified Psychoactive Substance Intoxication, Other or Unspecified Psychoactive Substance Delirium, Other or Unspecified Psychoactive Substance Dementia, Other or Unspecified Psychoactive Substance Delusional Disorder, Other or Unspecified Psychoactive Substance Hallucinosi

5 Disorder, Other or Unspecified Psychoactive Substance Anxiety Disorder, Other or Unspecified Psychoactive Substance Personality Disorder, Other or Unspecified Psychoactive Substance Organic Mental Disorder NOS, Delirium, Dementia, Organic Delusional Disorder, Organic Hallucinosi

10 Disorder, Organic Hallucinosi, Organic Mood Disorder, Organic Anxiety Disorder, Organic Personality Disorder, Organic Mental Disorder, Obsessive Compulsive Disorder, Post-traumatic Stress Disorder, Generalized Anxiety Disorder, Anxiety Disorder NOS, Body Dysmorphic Disorder, Hypochondriasis (or Hypochondriacal Neurosis), Somatization Disorder, Undifferentiated Somatoform Disorder, Somatoform Disorder NOS, Intermittent Explosive Disorder, Kleptomania, Pathological Gambling, Pyromania, Trichotillomania and Impulse Control Disorder NOS.

15

Additional examples of pathologic psychological conditions which may be treated as described in this invention include Schizophrenia, Catatonic, Subchronic, Schizophrenia, Catatonic, Chronic, Schizophrenia, Catatonic, Subchronic with Acute Exacerbation, Schizophrenia, Catatonic, Chronic with Acute Exacerbation, Schizophrenia, Catatonic, in

20 Remission, Schizophrenia, Catatonic, Unspecified, Schizophrenia, Disorganized, Chronic, Schizophrenia, Disorganized, Subchronic with Acute Exacerbation, Schizophrenia, Disorganized, Chronic with Acute Exacerbation, Schizophrenia, Disorganized, in Remission, Schizophrenia, Disorganized, Unspecified, Schizophrenia, Paranoid, Subchronic, Schizophrenia, Paranoid, Chronic, Schizophrenia, Paranoid, Subchronic with

25 Acute Exacerbation, Schizophrenia, Paranoid, Chronic with Acute Exacerbation, Schizophrenia, Paranoid, in Remission, Schizophrenia, Paranoid, Unspecified, Schizophrenia, Undifferentiated, Subchronic, Schizophrenia, Undifferentiated, Chronic, Schizophrenia, Undifferentiated, Subchronic with Acute Exacerbation, Schizophrenia, Undifferentiated, Chronic with Acute Exacerbation (295.94), Schizophrenia,

30 Undifferentiated, in Remission, Schizophrenia, Undifferentiated, Unspecified, Schizophrenia, Residual, Subchronic, Schizophrenia, Residual, Chronic, Schizophrenia,

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Residual, Subchronic with Acute Exacerbation, Schizophrenia, Residual, Chronic with
Acute Exacerbation, Schizophrenia, Residual, in Remission, Schizophrenia, Residual,
unspecified, Delusional (Paranoid) Disorder, Brief Reactive Psychosis, Schizophreniform
Disorder, Schizoaffective Disorder, induced Psychotic Disorder, Psychotic Disorder NOS
5 (Atypical Psychosis), Bipolar Disorder, Mixed, Severe, without Psychotic Features,
Bipolar Disorder, Manic, Severe, without Psychotic Features, Bipolar Disorder, Depressed,
Severe, without Psychotic Features, Bipolar Disorder, Mixed, with Psychotic Features,
Bipolar Disorder, Manic, with Psychotic Features, Bipolar Disorder, Depressed, with
Psychotic Features, Bipolar Disorder NOS, Major Depression, Single Episode, with
10 Psychotic Features, Major Depression, Recurrent with Psychotic Features Personality
Disorders, Paranoid Personality Disorders, Schizoid, Personality Disorders, Schizotypal,
Personality Disorders, Antisocial, Personality Disorders, Borderline.

Anxiety disorders which may be treated in accordance with this invention include, but are
15 not limited to, Anxiety Disorders, Panic Disorder), Panic Disorder with Agoraphobia,
Panic Disorder without Agoraphobia, Agoraphobia without History of Panic Disorders,
Social Phobia, Simple Phobia, Organic Anxiety Disorder, Psychoactive Substance Anxiety
Disorder, Separation Anxiety Disorder, Avoidant Disorder of Childhood or Adolescence,
and Overanxious Disorder.

20 Moderate Mental Retardation; Severe Mental Retardation; Profound Mental Retardation;
Autistic Disorder; Attention-Deficit Hyperactivity Disorder; Pervasive Development
Disorder NOS; Conduct Disorder, Group Type; Conduct Disorder, Solitary Aggressive
Type; Tourette's Disorder; Primary Degenerative Dementia of the Alzheimer Type, Senile
25 Onset, with Delirium; Primary Degenerative Dementia of the Alzheimer Type, Senile
Onset, with Delusions; Primary Degenerative Dementia of the Alzheimer Type, Presenile
Onset; Schizophrenia, Catatonic, Subchronic; Schizophrenia, Catatonic, Chronic;
Schizophrenia, Catatonic, Subchronic with Acute Exacerbation; Schizophrenia, Catatonic,
Chronic with Acute Exacerbation; Schizophrenia, Catatonic, in Remission; Schizophrenia,
30 Catatonic, Unspecified; Schizophrenia, Disorganized, Subchronic; Schizophrenia,
Disorganized, Chronic; Schizophrenia, Disorganized, Subchronic with Acute

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- Exacerbation; Schizophrenia, Disorganized, Chronic with Acute Exacerbation; Schizophrenia, Disorganized, in Remission; Schizophrenia, Disorganized, Unspecified; Schizophrenia, Paranoid, Subchronic; Schizophrenia, Paranoid, Chronic; Schizophrenia, Paranoid, Subchronic with Acute Exacerbation; Schizophrenia, Paranoid, Chronic with
- 5 Acute Exacerbation; Schizophrenia, Paranoid, in Remission; Schizophrenia, Paranoid, Unspecified; Schizophrenia, Undifferentiated, Subchronic; Schizophrenia, Undifferentiated, Chronic; Schizophrenia, Undifferentiated, Subchronic with Acute Exacerbation; Schizophrenia, Undifferentiated, Chronic with Acute Exacerbation; Schizophrenia, Undifferentiated, in Remission; Schizophrenia, Undifferentiated,
- 10 Unspecified; Schizophrenia, Residual, Subchronic; Schizophrenia, Residual Chronic; Schizophrenia, Residual, Subchronic with Acute Exacerbation; Schizophrenia, Residual, Chronic with Acute Exacerbation; Schizophrenia, Residual, in Remission; Schizophrenia, Residual, Unspecified; Delusional (Paranoid) Disorder; Brief Reactive Psychosis; Schizophreniform Disorder; Schizoaffective Disorder; Induced Psychotic Disorder;
- 15 Psychotic Disorder NOS (Atypical Psychosis); Bipolar Disorder, Mixed, with Psychotic Features; Bipolar Disorder, Manic, with Psychotic Features; Bipolar Disorder, Depressed, with Psychotic Features; Bipolar Disorder NOS; Major Depression, Single Episode, or Recurrent with Psychotic Features; Personality Disorders, Paranoid; Personality Disorders, Schizoid; Personality Disorders, Schizotypal; Personality Disorders, Antisocial;
- 20 Personality Disorders, Borderline, Anxiety Disorders, Panic Disorder, Panic Disorder with Agoraphobia, Panic Disorder without Agoraphobia, Agoraphobia without History of Panic Disorders, Social Phobia, Simple Phobia, Obsessive Compulsive Disorder, Post-Traumatic Stress Disorder, Generalized Anxiety Disorder, Anxiety Disorder NOS, Organic Anxiety Disorder, Psychoactive Substance Anxiety Disorder, Separation Anxiety Disorder,
- 25 Avoidant Disorder of Childhood or Adolescence, and Overanxious Disorder.

- Psychotic conditions contemplated herein include Schizophrenia, Catatonic, Subchronic; Schizophrenia, Catatonic, Chronic; Schizophrenia, Catatonic, Subchronic with Acute Exacerbation; Schizophrenia, Catatonic, Chronic with Acute Exacerbation; Schizophrenia,
- 30 Catatonic, in Remission; Schizophrenia, Catatonic, Unspecified; Schizophrenia, Disorganized, Subchronic; Schizophrenia, Disorganized, Chronic; Schizophrenia,

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Disorganized, Subchronic with Acute Exacerbation; Schizophrenia, Disorganized, Chronic with Acute Exacerbation; Schizophrenia, Disorganized, in Remission; Schizophrenia, Disorganized, Unspecified; Schizophrenia, Paranoid, Subchronic; Schizophrenia, Paranoid, Chronic; Schizophrenia, Paranoid, Subchronic with Acute Exacerbation; Schizophrenia, 5 Paranoid, Chronic with Acute Exacerbation; Schizophrenia, Paranoid, in Remission; Schizophrenia, Paranoid, Unspecified; Schizophrenia, Undifferentiated, Subchronic; Schizophrenia, Undifferentiated, Chronic; Schizophrenia, Undifferentiated, Subchronic with Acute Exacerbation; Schizophrenia, Undifferentiated, Chronic with Acute Exacerbation; Schizophrenia, Undifferentiated, in Remission; Schizophrenia, 10 Undifferentiated, Unspecified; Schizophrenia, Residual, Subchronic; Schizophrenia, Residual, Chronic; Schizophrenia, Residual, Subchronic with Acute Exacerbation; Schizophrenia, Residual, Chronic with Acute Exacerbation; Schizophrenia, Residual, in Remission; Schizophrenia, Residual, Unspecified; Delusional (Paranoid) Disorder; Brief Reactive Psychosis; Schizophreniform Disorder; Schizoaffective Disorder; Induced 15 Psychotic Disorder; Psychotic Disorder NOS (Atypical Psychosis); Bipolar Disorder, Mixed, with Psychotic Features; Bipolar Disorder, Manic, with Psychotic Features; Bipolar Disorder, Depressed, with Psychotic Features; Bipolar Disorder NOS; Personality Disorders, Paranoid; Personality Disorders, Schizoid; Personality Disorders, Schizotypal; Personality Disorders, Antisocial; Personality Disorders, Borderline.

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Reference to cardiovascular disease includes strokes and any condition of the systemic vasculature and includes atherosclerosis, chronic heart failure and general heart disease.

Other conditions contemplated herein include but are not limited to Adult Respiratory 25 distress syndrome, A-Beta-Lipoproteinemia, A-V, A Beta-2-Microglobulin Amyloidosis, A-T, A1AD, A1AT, Aagenaes, Aarskog syndrome, Aarskog-Scott Syndrome, Aase-smith syndrome, Aase Syndrome, AAT, Abderhalden-Kaufmann-Lignac Syndrome, Abdominal Muscle Deficiency Syndrome, Abdominal Wall Defect, Abdominal Epilepsy, Abdominal Migraine, Abductor Spasmodic Dysphonia, Abductor Spastic Dysphonia, Abercrombie 30 Syndrome, blepharon-Macrostromia Syndrome, ABS, Absence of HPRT, Absence of Corpus Callosum Schinzel Typ, Absence Defect of Limbs Scalp and Skull, Absence of

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- Menstruation Primar, Absence of HGPRT, Absorptive Hyperoxaluria or Enteric, Abt-
 Letterer-Siwe Disease, ACADL, ACADM Deficiency, ACADM, ACADS,
 Acanthocytosis-Neurologic Disorder, Acanthocytosis, Acantholysis Bullosa, Acanthosis
 Nigricans, Acanthosis Bullosa, Acanthosis Nigricans With Insulin Resistance Type A,
 5 Acanthosis Nigricans With Insulin Resistance Type B, Acanthotic Nevus, Acatalasemia,
 Acatalasia, ACC, Accessory Atrioventricular Pathways, Accessory Atrioventricular
 Pathways, Acephaly, ACF with Cardiac Defects, Achalasia, Achard-Thiers Syndrome,
 ACHARD (Marfan variant), Achard's syndrome, Acholuric Jaundice, Achondrogenesis,
 Achondrogenesis Type IV, Achondrogenesis Type III, Achondroplasia, Achondroplasia
 10 Tarda, Achondroplastic Dwarfism, Achoo Syndrome, Achromat, Achromatope,
 Achromatopic, Achromatopsia, Achromic Nevi, Acid Ceramidase Deficiency, Acid
 Maltase Deficiency, Acid Beta-glucosidase Deficiency, Acidemia Methylmalonic,
 Acidemia Propionic, Acidemia with Episodic Ataxia and Weakness, Acidosis, Aclasis
 Tarsoepiphyseal, ACM, Acoustic Neurilemoma, Acoustic Neuroma, ACPS with Leg
 15 Hypoplasia, ACPS II, ACPS IV, ACPS III, Acquired Aphasia with Convulsive Disorder,
 Acquired Brown Syndrome, Acquired Epileptic Aphasia, Acquired Factor XIII Deficiency,
 Acquired Form of ACC (caused by infection while still in womb), Acquired
 Hyperoxaluria, Acquired Hypogammaglobulinemia, Acquired Immunodeficiency
 Syndrome (AIDS), Acquired Iron Overload, Acquired Lipodystrophy, Acquired Partial
 20 Lipodystrophy, Acquired Wandering Spleen, ACR, Acral Dysostosis with Facial and
 Genital Abnormalities, Acro Renal, Acrocallosal Syndrome Schinzel Type,
 Acrocephalosyndactyly, Acrocephalosyndactyly Type I, Acrocephalosyndactyly Type I
 Subtype I, Acrocephalopolysyndactyly Type II, Acrocephalopolysyndactyly Type III,
 Acrocephalopolysyndactyly Type IV, Acrocephalosyndactyly V (ACS5 or ACS V)
 25 Subtype I, Acrocephaly Skull Asymmetry and Mild Syndactyly, Acrocephaly,
 Acrochondrohyperplasia, Acrodermatitis Enteropathica, Acrodysostosis, Acrodystrophic
 Neuropathy, Acrofacial Dysostosis Nager Type, Acrofacial Dysostosis Postaxial Type,
 Acrofacial Dysostosis Type Genee-Wiedep, Acrogeria Familial, Acromegaly,
 Acromelalgia Hereditary, Acromesomelic Dysplasia, Acromesomelic Dwarfism,
 30 Acromicric Skeletal Dysplasia, Acromicric Dysplasia, Acroosteolysis with Osteoporosis
 and Changes in Skull and Mandible, Acroosteolysis, Acroparesthesia, ACS I, ACS Type

- II, ACS Type III, ACS, ACS3, ACTH Deficiency, Action Myoclonus, Acute Brachial Neuritis Syndrome, Acute Brachial Radiculitis Syndrome, Acute Cerebral Gaucher Disease, Acute Cholangitis, Acute Disseminated Encephalomyeloradiculopathy, Acute Disseminated Histiocytosis-X, Acute Hemorrhagic Polioencephalitis, Acute Idiopathic
- 5 Polyneuritis, Acute Immune-Mediation Polyneuritis, Acute Infantile Pelizaeus-Merzbacher Brain Sclerosis, Acute Intermittant Porphyria, Acute Porphyrias, Acute Sarcoidosis, Acute Shoulder Neuritis, Acute Toxic Epidermolysis, Acyl-CoA Dehydrogenase Deficiency Long-Chain, Acyl-CoA Dehydrogenase Deficiency Short-Chain, Acyl-CoA Dihydroxyacetone Acyltransferase, Acyl-coenzyme A Oxidase Deficiency, ADA, ADA
- 10 Deficiency, Adam Complex, Adamantiades-Behcet's Syndrome, Adamantinoma, Adams Oliver Syndrome, Adaptive Colitis, ADD combined type, ADD, Addison Disease with Cerebral Sclerosis, Addison's Anemia, Addison's Disease, Addison-Biermer Anemia, Addison-Schilder Disease, Addisonian Pernicious Anemia, Adducted Thumbs-Mental Retardation, Adductor Spasmodic Dysphonia, Adductor Spastic Dysphonia, Adenoma
- 15 Associated Virilism of Older Women, Adenomatosis of the Colon and Rectum, Adenomatous polyposis of the Colon, Adenomatous Polyposis Familial, Adenosine Deaminase Deficiency, Adenylosuccinase deficiency, ADHD predominantly hyperactive-impulsive type, ADHD predominantly inattentive type, ADHD, Adhesive Arachnoiditis, Adie Syndrome, Adie's Syndrome, Adie's Tonic Pupil, Adie's Pupil, Adipogenital
- 20 Retinitis Pigmentosa Polydactyly, Adipogenital-Retinitis Pigmentosa Syndrome, Adiposa Dolorosa, Adiposis Dolorosa, Adiposogenital Dystrophy, Adolescent Cystinosis, ADPKD, Adrenal Cortex Adenoma, Adrenal Disease, Adrenal Hyperfunction resulting from Pituitary ACTH Excess, Adrenal Hypoplasia, Adrenal Insufficiency, Adrenal Neoplasm, Adrenal Virilism, Adreno-Retinitis Pigmentosa-Polydactyly Syndrome, Adrenocortical
- 25 Insufficiency, Adrenocortical Hypofunction, Adrenocorticotrophic Hormone Deficiency Isolated, Adrenogenital Syndrome, Adrenoleukodystrophy, Adrenomyeloneuropathy, Adreno-Retinitis Pigmentosa-Polydactyly Syndrome, Adult Cystinosis, Adult Dermatomyositis, Adult Hypophosphatasia, Adult Macula Lutea Retinae Degeneration, Adult Onset ALD, Adult-Onset Ceroidosis, Adult Onset Medullary Cystic Disease, Adult
- 30 Onset Pernicious Anemia, Adult Onset Schindler Disease, Adult-Onset Subacute Necrotizing Encephalomyelopathy, Adult Polycystic Kidney Disease, Adult Onset

Medullary Cystic Disease, Adynlosuccinate Lyase Deficiency, AE, AEC Syndrome, AFD, Afibrinogenemia, African Siderosis, AGA, Aganglionic Megacolon, Age Related Macular Degeneration, Agenesis of Commissura Magna Cerebri, Agenesis of Corpus Callosum, Agenesis of Corpus Callosum-Infantile Spasms-Ocular Anomalies, Agenesis of Corpus

5 Callosum and Chorioretinal Abnormality, Agenesis of Corpus Callosum-Chorioretinitis Abnormality, Aggressive mastocytosis, Agnosis Primary, AGR Triad, AGU, Agyria, Agyria-pachygyria-band spectrum, AHC, AHD, AHDS, AHF Deficiency, AHG Deficiency, AHO, Ahumada Del Castillo, Aicardi Syndrome, AIED, AIMP, AIP, AIS, Akinetic Seizure, ALA-D Porphyria, Alactasia, Alagille Syndrome, Aland Island Eye Disease (X-

10 Linked), Alaninuria, Albers-Schonberg Disease, Albinism, Albinismus, Albinoidism, Albright Hereditary Osteodystrophy, Alcaptonuria, Alcohol-Related Birth Defects, Alcoholic Embryopathy, Ald, ALD, ALD, Aldosterone, Aldosteronism With Normal Blood Pressure, Aldrich Syndrome, Alexander's Disease, Alexanders Disease, Algodystrophy, Algoneurodystrophy, Alkaptonuria, Alkaptonuric Ochronosis, Alkyl

15 DHAP synthase deficiency, Allan-Herndon-Dudley Syndrome, Allan-Herndon Syndrome, Allan-Herndon-Dudley Mental Retardation, Allergic Granulomatous Antitis, Allergic Granulomatous Angiitis of Cronkhite-Canada, Alobar Holoprosencephaly, Alopecia Areata, Alopecia Celsi, Alopecia Cicatrisata, Alopecia Circumscripta, Alopecia-Poliosis-Uveitis-Vitiligo-Deafness-Cutaneous-Uveo-O, Alopecia Seminuniversalis, Alopecia

20 Totalis, Alopecia Universalis, Alpers Disease, Alpers Diffuse Degeneration of Cerebral Gray Matter with Hepatic Cirrhosis, Alpers Progressive Infantile Poliodystrophy, Alpha-1-Antitrypsin Deficiency, Alpha-1 4 Glucosidase Deficiency, Alpha-Galactosidase A Deficiency, Alpha-Galactosidase B Deficiency, Alpha High-Density Lipoprotein Deficiency, Alpha-L-Fucosidase Deficiency Fucosidosis Type 3, Alpha-GalNAc Deficiency

25 Schindler Type, Alphalipoproteinemia, Alpha Mannosidosis, Alpha-N-Acetylgalactosaminidase Deficiency Schindler Type, Alpha-NAGA Deficiency Schindler Type, Alpha-Neuraminidase Deficiency, Alpha-Thalassemia/mental retardation syndrome non-deletion type, Alphaschindler, Alport Syndrome, ALS, Alstroem's Syndrome, Alstroem, Alstrom Syndrome, Alternating Hemiplegia Syndrome, Alternating Hemiplegia

30 of Childhood, Alzheimer's Disease, Amaurotic Familial Idiocy, Amaurotic Familial Idiocy Adult, Amaurotic Familial Infantile Idiocy, Ambiguous Genitalia, AMC, AMD,

- Ameloblastoma, Amelogenesis Imperfecta, Amenorrhea-Galactorrhea Nonpuerperal, Amenorrhea-Galactorrhea-FSH Decrease Syndrome, Amenorrhea, Amino Acid Disorders, Aminoaciduria-Osteomalacia-Hyperphosphaturia Syndrome, AMN, Amniocentesis, Amniotic Bands, Amniotic Band Syndrome, Amniotic Band Disruption Complex,
- 5 Amniotic Band Sequence, Amniotic Rupture Sequence, Amputation Congenital, AMS, Amsterdam Dwarf Syndrome de Lange, Amylo-1 6-Glucosidase Deficiency, Amyloid Arthropathy of Chronic Hemodialysis, Amyloid Corneal Dystrophy, Amyloid Polyneuropathy, Amyloidosis, Amyloidosis of Familial Mediterranean Fever, Amylopectinosis, Amyoplasia Congenita, Amyotrophic Lateral Sclerosis, Amyotrophic
- 10 Lateral Sclerosis, Amyotrophic Lateral Sclerosis-Polyglucosan Bodies, AN, AN 1, AN 2, Anal Atresia, Anal Membrane, Anal Rectal Malformations, Anal Stenosis, Analine 60 Amyloidosis, Analphalipoproteinemia, Analrectal, Analrectal, Anaplastic Astrocytoma, Andersen Disease, Anderson-Fabry Disease, Andersen Glycogenosis, Anderson-Warburg Syndrome, Andre Syndrome, Andre Syndrome Type II, Androgen Insensitivity, Androgen
- 15 Insensitivity Syndrome Partial, Androgen Insensitivity Syndrome Partial, Androgenic Steroids, Anemia Autoimmune Hemolytic, Anemia Blackfan Diamond, Anemia, Congenital, Triphalangeal Thumb Syndrome, Anemia Hemolytic-Cold Antibody, Anemia Hemolytic with PGK Deficiency, Anemia Pernicious, Anencephaly, Angelman Syndrome, Angio-Osteohypertrophy Syndrome, Angiofollicular Lymph Node Hyperplasia,
- 20 Angiohemophilia, Angiokeratoma Corporis, Angiokeratoma Corporis Diffusum, Angiokeratoma Diffuse, Angiomatosis Retina, Angiomatous Lymphoid, Angioneurotic Edema Hereditary, Anhidrotic Ectodermal Dysplasia, Anhidrotic X-Linked Ectodermal Dysplasias, Aniridia, Aniridia-Ambiguous Genitalia-Mental Retardation, Aniridia Associated with Mental Retardation, Aniridia-Cerebellar Ataxia-Mental Deficiency,
- 25 Aniridia Partial-Cerebellar Ataxia-Mental Retardation, Aniridia Partial-Cerebellar Ataxia-Oligophrenia, Aniridia Type I, Aniridia Type II, Aniridia-Wilms' Tumor Association, Aniridia-Wilms' Tumor-Gonadoblastoma, Ankyloblepharon-Ectodermal Defects-Cleft Lip/Palate, Ankylosing Spondylitis, Annular groves, Anodontia, Anodontia Vera, Anomalous Trichromasy, Anomalous Dysplasia of Dentin, Coronal Dentin Dysplasia,
- 30 Anomic Aphasia, Anophthalmia, Anorectal, Anorectal Malformations, Anosmia, Anterior Bowing of the Legs with Dwarfism, Anterior Membrane Corneal Dystrophy, Anti-

Convulsant Syndrome, Anti-Epstein-Barr Virus Nuclear Antigen (EBNA) Antibody Deficiency, Antibody Deficiency, Antibody Deficiency with near normal Immunoglobulins, Antihemophilic Factor Deficiency, Antihemophilic Globulin Deficiency, Antiphospholipid Syndrome, Antiphospholipid Antibody Syndrome, 5 Antithrombin III Deficiency, Antithrombin III Deficiency Classical (Type I), Antitrypsin Deficiency, Antley-Bixler Syndrome, Antoni's Palsy, Anxietas Tibialis, Aorta Arch Syndrome, Aortic and Mitral Atresia with Hypoplastic Left Heart Syndrome, Aortic Stenosis, Aparoschisis, APC, APECED Syndrome, Apert Syndrome, Aperts, Aphasia, Aplasia Axialis Extracorticales Congenital, Aplasia Cutis Congenita, Aplasia Cutis 10 Congenita with Terminal Transverse Limb Defects, Aplastic Anemia, Aplastic Anemia with Congenital Anomalies, APLS, Apnea, Appalachian Type Amyloidosis, Apple Peel Syndrome, Apraxia, Apraxia Buccofacial, Apraxia Constructional, Apraxia Ideational, Apraxia Ideokinetic, Apraxia Ideomotor, Apraxia Motor, Apraxia Oculomotor, APS, Arachnitis, Arachnodactyly Contractural Beals Type, Arachnodactyly, Arachnoid Cysts, 15 Arachnoiditis Ossificans, Arachnoiditis, Aran-Duchenne, Aran-Duchenne Muscular Atrophy, Aregenerative Anemia, Arginase Deficiency, Argininemia, Arginino Succinase Deficiency, Argininosuccinase Deficiency, Argininosuccinate Lyase Deficiency, Argininosuccinic Acid Lyase-ASL, Argininosuccinic Acid Synthetase Deficiency, Argininosuccinic Aciduria, Argonz-Del Castillo Syndrome, Arhinencephaly, Armenian 20 Syndrome, Arnold-Chiari Malformation, Arnold-Chiari Syndrome, ARPKD, Arrhythmic Myoclonus, Arrhythmogenic Right Ventricular Dysplasia, Arteriohepatic Dysplasia, Arteriovenous Malformation, Arteriovenous Malformation of the Brain, Arteritis Giant Cell, Arthritis, Arthritis Urethritica, Arthro-Dento-Osteodysplasia, Arthro-Ophthalmopathy, Arthrochhalasis Multiplex Congenita, Arthrogryposis Multiplex 25 Congenita, Arthrogryposis Multiplex Congenita, Distal, Type IIA, ARVD, Arylsulfatase-B Deficiency, AS, ASA Deficiency, Ascending Paralysis, ASD, Atrioseptal Defects, ASH, Ashermans Syndrome, Ashkenazi Type Amyloidosis, ASL Deficiency, Aspartylglucosaminuria, Aspartylglycosaminuria, Asperger's Syndrome, Asperger's Type Autism, Asphyxiating Thoracic Dysplasia, Asplenia Syndrome, ASS Deficiency, Asthma, 30 Astrocytoma Grade I (Benign), Astrocytoma Grade II (Benign), Asymmetric Crying Facies with Cardiac Defects, Asymmetrical septal hypertrophy, Asymptomatic Callosal

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- Agensis, AT, AT III Deficiency, AT III Variant IA, AT III Variant Ib, AT 3, Ataxia, Ataxia Telangiectasia, Ataxia with Lactic Acidosis Type II, Ataxia Cerebral Palsy, Ataxiadynamia, Ataxiophemia, ATD, Athetoid Cerebral Palsy, Atopic Eczema, Atresia of Esophagus with or without Tracheoesophageal Fistula, Atrial Septal Defects, Atrial Septal
- 5 Defect Primum, Atrial and Septal and Small Ventricular Septal Defect, Atrial Flutter, Atrial Fibrillation, Atriодigital Dysplasia, Atrioseptal Defects, Atrioventricular Block, Atrioventricular Canal Defect, Atrioventricular Septal Defect, Atrophia Bulborum Hereditaria, Atrophic Beriberi, Atrophy Olivopontocerebellar, Attention Deficit Disorder, Attention Deficit Hyperactivity Disorder, Attenuated Adenomatous Polyposis Coli,
- 10 Atypical Amyloidosis, Atypical Hyperphenylalaninemia, Auditory Canal Atresia, Auriculotemporal Syndrome, Autism, Autism Asperger's Type, Autism Dementia Ataxia and Loss of Purposeful Hand Use, Autism Infantile Autism, Autoimmune Addison's Disease, Autoimmune Hemolytic Anemia, Autoimmune Hepatitis, Autoimmune-Polyendocrinopathy-Candidias, Autoimmune Polyglandular Disease Type I, Autosomal
- 15 Dominant Albinism, Autosomal Dominant Compelling Heliophthalmic Outburst Syndrome, Autosomal Dominant Desmin Distal myopathy with Late Onset, Autosomal Dominant EDS, Autosomal Dominant Emery-Dreifuss Muscular Dystrophy, Autosomal Dominant Keratoconus, Autosomal Dominant Pelizaeus-Merzbacher Brain Sclerosis, Autosomal Dominant Polycystic Kidney Disease, Autosomal Dominant Spinocerebellar
- 20 Degeneration, Autosomal Recessive Agammaglobulinemia, Autosomal Recessive Centronuclear myopathy, Autosomal Recessive Conradi-Hunermann Syndrome, Autosomal Recessive EDS, Autosomal Recessive Emery-Dreifuss Muscular Dystrophy, Autosomal Recessive Forms of Ocular Albinism, Autosomal Recessive Inheritance Agensis of Corpus Callosum, Autosomal Recessive Keratoconus, Autosomal Recessive
- 25 Polycystic Kidney Disease, Autosomal Recessive Severe Combined Immunodeficiency, AV, AVM, AVSD, AWTa, Axilla Abscess, Axonal Neuropathy Giant, gan.htm Azorean Neurologic Disease, B-K Mole Syndrome, Babinski-Froelich Syndrome, BADS, Baillarger's Syndrome, Balkan Disease, Baller-Gerold Syndrome, Ballooning Mitral Valve, Balo Disease Concentric Sclerosis, Baltic Myoclonus Epilepsy, Bannayan-Zonana
- 30 syndrome (BZS), Bannayan-Riley-Ruvalcaba syndrome, Banti's Disease, Bardet-Biedl Syndrome, Bare Lymphocyte Syndrome, Barlow's syndrome, Barraquer-Simons Disease,

Barrett Esophagus, Barrett Ulcer, Barth Syndrome, Bartter's Syndrome, Basal Cell Nevus Syndrome, Basedow Disease, Bassen-Kornzweig Syndrome, Batten Disease, Batten-Mayou Syndrome, Batten-Spielmeyer-Vogt's Disease, Batten Turner Syndrome, Batten Turner Type Congenital myopathy, Batten-Vogt Syndrome, BBB Syndrome, BBB Syndrome (Opitz), BBB Syndrome, BBBG Syndrome, BCKD Deficiency, BD, BDLS, BE, 5 Beals Syndrome, Beals Syndrome, Beals-Hecht Syndrome, Bean Syndrome, BEB, Bechterew Syndrome, Becker Disease, Becker Muscular Dystrophy, Becker Nevus, Beckwith Wiedemann Syndrome, Beckwith-Syndrome, Begnez-Cesar's Syndrome, Behcet's syndrome, Behcet's Disease, Behr 1, Behr 2, Bell's Palsy, Benign Acanthosis 10 Nigricans, Benign Astrocytoma, Benign Cranial Nerve Tumors, Benign Cystinosis, Benign Essential Blepharospasm, Benign Essential Tremor, Benign Familial Hematuria, Benign Focal Amyotrophy, Benign Focal Amyotrophy of ALS, Benign Hydrocephalus, Benign Hypermobility Syndrome, Benign Keratosis Nigricans, Benign Paroxysmal Peritonitis, Benign Recurrent Hematuria, Benign Recurrent Intrahepatic Cholestasis, Benign Spinal 15 Muscular Atrophy with Hypertrophy of the Calves, Benign Symmetrical Lipomatosis, Benign Tumors of the Central Nervous System, Berardinelli-Seip Syndrome, Berger's Disease, Beriberi, Berman Syndrome, Bernard-Horner Syndrome, Bernard-Soulier Syndrome, Besnier Prurigo, Best Disease, Beta-Alanine-Pyruvate Aminotransferase, Beta-Galactosidase Deficiency Morquio Syndrome, Beta-Glucuronidase Deficiency, Beta 20 Oxidation Defects, Beta Thalassemia Major, Beta Thalassemia Minor, Betalipoprotein Deficiency, Bethlem myopathy, Beuren Syndrome, BH4 Deficiency, Biber-Haab-Dimmer Corneal Dystrophy, Bicuspid Aortic Valve, Biedl-Bardet, Bifid Cranium, Bifunctional Enzyme Deficiency, Bilateral Acoustic Neurofibromatosis, Bilateral Acoustic Neuroma, Bilateral Right-Sidedness Sequence, Bilateral Renal Agenesis, Bilateral Temporal Lobe 25 Disorder, Bilious Attacks, Bilirubin Glucuronosyltransferase Deficiency Type I, Binder Syndrome, Binswanger's Disease, Binswanger's Encephalopathy, Biotinidase deficiency, Bird-Headed Dwarfism Seckel Type, Birth Defects, Birthmark, Bitemporal Forceps Marks Syndrome, Biventricular Fibrosis, Bjornstad Syndrome, B-K Mole Syndrome, Black Locks-Albinism-Deafness of Sensoneural Type (BADs), Blackfan-Diamond Anemia, 30 Blennorrheal Idiopathic Arthritis, Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome, Blepharospasm, Blepharospasm Benign Essential, Blepharospasm

- Oromandibular Dystonia, Blessig Cysts, BLFS, Blindness, Bloch-Siemens Incontinentia Pigmenti Melanoblastosis Cutis Linearis, Bloch-Siemens-Sulzberger Syndrome, Bloch-Sulzberger Syndrome, Blood types, Blood type A, Blood type B, Blood type AB, Blood type O, Bloom Syndrome, Bloom-Torre-Mackacek Syndrome, Blue Rubber Bleb Nevus,
- 5 Blue Baby, Blue Diaper Syndrome, BMD, BOD, BOFS, Bone Tumor-Epidermoid Cyst-Polyposis, Bonnet-Dechaume-Blanc Syndrome, Bonnevie-Ulrich Syndrome, Book Syndrome, BOR Syndrome, BORJ, Borjeson Syndrome, Borjeson-Forssman-Lehmann Syndrome, Bowen Syndrome, Bowen-Conradi Syndrome, Bowen-Conradi Hutterite, Bowen-Conradi Type Hutterite Syndrome, Bowman's Layer, BPEI, BPES, Brachial
- 10 Neuritis, Brachial Neuritis Syndrome, Brachial Plexus Neuritis, Brachial-Plexus-Neuropathy, Brachiocephalic Ischemia, Brachmann-de Lange Syndrome, Brachycephaly, Brachymorphic Type Congenital, Bradycardia, Brain Tumors, Brain Tumors Benign, Brain Tumors Malignant, Branched Chain Alpha-Ketoacid Dehydrogenase Deficiency, Branched Chain Ketonuria I, Brancher Deficiency, Branchio-Oculo-Facial Syndrome, Branchio-Oto-
- 15 Renal Dysplasia, Branchio-Oto-Renal Syndrome, Branchiooculofacial Syndrome, Branchiootic Syndrome, Brandt Syndrome, Brandywine Type Dentinogenesis Imperfecta, Brandywine type Dentinogenesis Imperfecta, Breast Cancer, BRIC Syndrome, Brittle Bone Disease, Broad Beta Disease, Broad Thumb Syndrome, Broad Thumbs and Great Toes Characteristic Facies and Mental Retardation, Broad Thumb-Hallux, Broca's
- 20 Aphasia, Brocq-Duhring Disease, Bronze Diabetes, Bronze Schilder's Disease, Brown Albinism, Brown Enamel Hereditary, Brown-Sequard Syndrome, Brown Syndrome, BRRS, Brueghel Syndrome, Bruton's Agammaglobulinemia Common, BS, BSS, Buchanan's Syndrome, Budd's Syndrome, Budd-Chiari Syndrome, Buerger-Gruetz Syndrome, Bulbospinal Muscular Atrophy-X-linked, Bulldog Syndrome, Bullosa
- 25 Hereditaria, Bullous CIE, Bullous Congenital Ichthyosiform Erythroderma, Bullous Ichthyosis, Bullous Pemphigoid, Burkitt's Lymphoma, Burkitt's Lymphoma African type, Burkitt's Lymphoma Non-african type, BWS, Byler's Disease, C Syndrome, C1 Esterase Inhibitor Dysfunction Type II Angioedema, C1-INH, C1 Esterase Inhibitor Deficiency Type I Angioedema, C1NH, Cacchi-Ricci Disease, CAD, CADASIL, CAH, Calcaneal
- 30 Valgus, Calcaneovalgus, Calcium Pyrophosphate Dihydrate Deposits, Callosal Agenesis and Ocular Abnormalities, Calves-Hypertrophy of Spinal Muscular Atrophy, Campomelic

- Dysplasia, Campomelic Dwarfism, Campomelic Syndrome, Camptodactyly-Cleft Palate-Clubfoot, Camptodactyly-Limited Jaw Excursion, Camptomelic Dwarfism, Camptomelic Syndrome, Camptomelic Syndrome Long-Limb Type, Camurati-Engelmann Disease, Canada-Cronkhite Disease, Canavan disease, Canavan's Disease Included, Canavan's
- 5 Leukodystrophy, Cancer, Cancer Family Syndrome Lynch Type, Cantrell Syndrome, Cantrell-Haller-Ravich Syndrome, Cantrell Pentalogy, Carbamyl Phosphate Synthetase Deficiency, Carbohydrate Deficient Glycoprotein Syndrome, Carbohydrate-Deficient Glycoprotein Syndrome Type Ia, Carbohydrate-Induced Hyperlipemia, Carbohydrate Intolerance of Glucose Galactose, Carbon Dioxide Acidosis, Carboxylase Deficiency
- 10 Multiple, Cardiac-Limb Syndrome, Cardio-auditory Syndrome, Cardioauditory Syndrome of Jervell and Lange-Nielsen, Cardiocutaneous Syndrome, Cardio-facial-cutaneous syndrome, Cardiofacial Syndrome Cayler Type, Cardiomegalia Glycogenica Diffusa, Cardiomyopathic Lentiginosis, Cardio myopathy, Cardio myopathy Associated with Desmin Storage myopathy, Cardio myopathy Due to Desmin Defect, Cardio myopathy-
- 15 Neutropenia Syndrome, Cardio myopathy-Neutropenia Syndrome Lethal Infantile Cardio myopathy, Cardiopathic Amyloidosis, Cardiospasm, Cardocardiac Syndrome, Carnitine-Acylcarnitine Translocase Deficiency, Carnitine Deficiency and Disorders, Carnitine Deficiency Primary, Carnitine Deficiency Secondary, Carnitine Deficiency Secondary to MCAD Deficiency, Carnitine Deficiency Syndrome, Carnitine Palmitoyl Transferase I &
- 20 II (CPT I & II), Carnitine Palmitoyltransferase Deficiency, Carnitine Palmitoyltransferase Deficiency Type 1, Carnitine Palmitoyltransferase Deficiency Type 2 benign classical muscular form included severe infantile form included, Carnitine Transport Defect (Primary Carnitine Deficiency), Carnosinase Deficiency, Carnosinemia, Caroli Disease, Carpenter syndrome, Carpenter's, Cartilage-Hair Hypoplasia, Castleman's Disease,
- 25 Castleman's Disease Hyaline Vascular Type, Castleman's Disease Plasma Cell Type, Castleman Tumor, Cat Eye Syndrome, Cat's Cry Syndrome, Catalayse deficiency, Cataract-Dental Syndrome, Cataract X-Linked with Hutchinsonian Teeth, Catecholamine hormones, Catel-Manzke Syndrome, Catel-Manzke Type Palatodigital Syndrome, Caudal Dysplasia, Caudal Dysplasia Sequence, Caudal Regression Syndrome, Causalgia
- 30 Syndrome Major, Cavernomas, Cavernous Angioma, Cavernous Hemangioma, Cavernous Lymphangioma, Cavernous Malformations, Cayler Syndrome, Cazenave's Vitiligo,

- CBGD, CBPS, CCA, CCD, CCHS, CCM Syndrome, CCMS, CCO, CD, CDG1a, CDG1A, CDGS Type Ia, CDGS, CDI, CdLS, Celiac Disease, Celiac sprue, Celiac Sprue-Dermatitis, Cellular Immunodeficiency with Purine Nucleoside Phosphorylase Deficiency, Celsus' Vitiligo, Central Apnea, Central Core Disease, Central Diabetes Insipidus, Central
- 5 Form Neurofibromatosis, Central Hypoventilation, Central Sleep Apnea, Centrifugal Lipodystrophy, Centronuclear myopathy, CEP, Cephalocele, Cephalothoracic Lipodystrophy, Ceramide Trihexosidase Deficiency, Cerebellar Agenesis, Cerebellar Aplasia, Cerebellar Hemiagenesis, Cerebellar Hypoplasia, Cerebellar Vermis Aplasia, Cerebellar Vermis Agenesis-Hypernea-Episodic Eye Moves-Ataxia-Retardation,
- 10 Cerebellar Syndrome, Cerebellarparenchymal Disorder IV, Cerebellomedullary Malformation Syndrome, Cerebello-Oculocutaneous Telangiectasia, Cerebelloparenchymal Disorder IV Familial, Cerebellopontine Angle Tumor, Cerebral Arachnoiditis, Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukodystrophy, Cerebral Beriberi, Cerebral Diplegia, Cerebral Gigantism, Cerebral
- 15 Malformations Vascular, Cerebral Palsy, Cerebro-Oculorenal Dystrophy, Cerebro-Oculo-Facio-Skeletal Syndrome, Cerebrocostomandibular syndrome, Cerebrohepatorenal Syndrome, Cerebromacular Degeneration, Cerebromuscular Dystrophy Fukuyama Type, Cerebroocular Dysgenesis, Cerebroocular Dysplasia-Muscular Dystrophy Syndrome, Cerebrooculofacioskeletal Syndrome, Cerebroretinal Arteriovenous Aneurysm,
- 20 Cerebroside Lipidosis, Cerebrosidosis, Cerebrotendinous Xanthomatosis, Cerebrovascular Ferrocalcinosis, Ceroid-Lipofuscinosis Adult form, Cervical Dystonia, Cervical Dystonia, Cervico-Oculo-Acoustic Syndrome, Cervical Spinal Stenosis, Cervical Vertebral Fusion, CES, CF, CFC syndrome, CFIDS, CFND, CGD, CGF, Chalasodermia Generalized, Chanarin Dorfman Disease, Chanarin Dorfman Syndrome, Chanarin Dorfman Ichthyosis
- 25 Syndrome, Chandler's Syndrome, Charcot's Disease, Charcot-Marie-Tooth, Charcot-Marie-Tooth Disease, Charcot-Marie-Tooth Disease Variant, Charcot-Marie-Tooth-Roussy-Levy Disease, CHARGE Association, Charge Syndrome, CHARGE Syndrome, Chaund's Ectodermal Dysplasias, Chediak-Higashi Syndrome, Chediak-Steinbrinck-Higashi Syndrome, Cheilitis Granulomatosa, Cheiloschisis, Chemke Syndrome, Cheney
- 30 Syndrome, Cherry Red Spot and Myoclonus Syndrome, CHF, CHH, Chiari's Disease, Chiari Malformation I, Chiari Malformation, Chiari Type I (Chiari Malformation I), Chiari

- 47 -

- Type II (Chiari Malformation II), Chiari I Syndrome, Chiari-Budd Syndrome, Chiari-Frommel Syndrome, Chiari Malformation II, CHILD Syndrome, CHILD Ichthyosis Syndrome, CHILD Syndrome Ichthyosis, Childhood Adrenoleukodystrophy, Childhood Dermatomyositis, Childhood-onset Dystonia, Childhood Cyclic Vomiting, Childhood
- 5 Giant Axonal Neuropathy, Childhood Hypophosphatasia, Childhood Muscular Dystrophy, CHN, Cholestasis, Cholestasis Hereditary Norwegian Type, Cholestasis Intrahepatic, Cholestasis Neonatal, Cholestasis of Oral Contraceptive Users, Cholestasis with Peripheral Pulmonary Stenosis, Cholestasis of Pregnancy, Cholesterol Desmolase Deficiency, Chondrodysplasia Punctata, Chondrodystrophia Calcificans Congenita, Chondrodystrophia
- 10 Fetalis, Chondrodystrophic Myotonia, Chondrodystrophy, Chondrodystrophy with Clubfeet, Chondrodystrophy Epiphyseal, Chondrodystrophy Hyperplastic Form, Chondroectodermal Dysplasias, Chondrogenesis Imperfecta, Chondrohystrophia, Chondroosteodystrophy, Choreoacanthocytosis, Chorionic Villi Sampling, Choriorretinal Anomalies, Choriorretinal Anomalies with ACC, Choriorretinal Coloboma-Joubert
- 15 Syndrome, Choroidal Sclerosis, Choroideremia, Chotzen Syndrome, Christ-Siemens-Touraine Syndrome, Christ-Siemans-Touraine Syndrome, Christmas Disease, Christmas Tree Syndrome, Chromosome 3 Deletion of Distal 3p, Chromosome 3 Distal 3p Monosomy, Chromosome 3-Distal 3q2 Duplication, Chromosome 3-Distal 3q2 Trisomy, Chromosome 3 Monosomy 3p2, Chromosome 3q Partial Duplication Syndrome,
- 20 Chromosome 3q, Partial Trisomy Syndrome, Chromosome 3-Trisomy 3q2, Chromosome 4 Deletion 4q31-qter Syndrome, Chromosome 4 Deletion 4q32-qter Syndrome, Chromosome 4 Deletion 4q33-qter Syndrome, Chromosome 4 Long Arm Deletion, Chromosome 4 Long Arm Deletion, Chromosome 4 Monosomy 4q, Chromosome 4-Monosomy 4q, Chromosome 4 Monosomy Distal 4q, Chromosome 4 Partial Deletion 4p,
- 25 Chromosome 4, Partial Deletion of the Short Arm, Chromosome 4 Partial Monosomy of Distal 4q, Chromosome 4 Partial Monosomy 4p, Chromosome 4 Partial Trisomy 4 (q25-qter), Chromosome 4 Partial Trisomy 4 (q26 or q27-qter), Chromosome 4 Partial Trisomy 4 (q31 or 32-qter), Chromosome 4 Partial Trisomy 4p, Chromosome 4 Partial Trisomies 4q2 and 4q3, Chromosome 4 Partial Trisomy Distal 4, Chromosome 4 Ring, Chromosome
- 30 4 4q Terminal Deletion Syndrome, Chromosome 4q- Syndrome, Chromosome 4q-Syndrome, Chromosome 4 Trisomy 4, Chromosome 4 Trisomy 4p, Chromosome 4 XY/47

XXY (Mosaic), Chromosome 5 Monosomy 5p, Chromosome 5, Partial Deletion of the Short Arm Syndrome, Chromosome 5 Trisomy 5p, Chromosome 5 Trisomy 5p Complete (5p11-pter), Chromosome 5 Trisomy 5p Partial (5p13 or 14-pter), Chromosome 5p-Syndrome, Chromosome 6 Partial Trisomy 6q, Chromosome 6 Ring, Chromosome 6 Trisomy 6q2, Chromosome 7 Monosomy 7p2, Chromosome 7 Partial Deletion of Short Arm (7p2-), Chromosome 7 Terminal 7p Deletion [del (7) (p21-p22)], Chromosome 8 Monosomy 8p2, Chromosome 8 Monosomy 8p21-pter, Chromosome 8 Partial Deletion (short arm), Chromosome 8 Partial Monosomy 8p2, Chromosome 9 Complete Trisomy 9P, Chromosome 9 Partial Deletion of Short Arm, Chromosome 9 Partial Monosomy 9p, Chromosome 9 Partial Monosomy 9p22, Chromosome 9 Partial Monosomy 9p22-pter, Chromosome 9 Partial Trisomy 9P Included, Chromosome 9 Ring, Chromosome 9 Tetrasomy 9p, Chromosome 9 Tetrasomy 9p Mosaicism, Chromosome 9 Trisomy 9p (Multiple Variants), Chromosome 9 Trisomy 9 (pter-p21 to q32) Included, Chromosome 9 Trisomy Mosaic, Chromosome 9 Trisomy Mosaic, Chromosome 10 Distal Trisomy 10q, Chromosome 10 Monosomy, Chromosome 10 Monosomy 10p, Chromosome 10, Partial Deletion (short arm), Chromosome 10, 10p- Partial, Chromosome 10 Partial Trisomy 10q24-qter, Chromosome 10 Trisomy 10q2, Partial Monosomy of Long Arm of Chromosome 11, Chromosome 11 Partial Monosomy 11q, Chromosome 11 Partial Trisomy, Chromosome 11 Partial Trisomy 11q13-qter, Chromosome 11 Partial Trisomy 11q21-qter, Chromosome 11 Partial Trisomy 11q23-qter, Chromosome 11q, Partial Trisomy, Chromosome 12 Isochromosome 12p Mosaic, Chromosome 13 Partial Monosomy 13q, Chromosome 13, Partial Monosomy of the Long Arm, Chromosome 14 Ring, Chromosome 14 Trisomy, Chromosome 15 Distal Trisomy 15q, Chromosome 15, Chromosome 15 Ring, Chromosome 15 Trisomy 15q2, Chromosome 15q, Partial Duplication Syndrome, Chromosome 17 Interstitial Deletion 17p, Chromosome 18 Long Arm Deletion Syndrome, Chromosome 18 Monosomy 18p, Chromosome 18 Monosomy 18Q, Chromosome 18 Ring, Chromosome 18 Tetrasomy 18p, Chromosome 18q-Syndrome, Chromosome 21 Mosaic 21 Syndrome, Chromosome 21 Ring, Chromosome 21 Translocation 21 Syndrome, Chromosome 22 Inverted Duplication (22pter-22q11), Chromosome 22 Partial Trisomy (22pter-22q11), Chromosome 22 Ring, Chromosome 22 Trisomy Mosaic, Chromosome 48 XXYY, Chromosome 48 XXXY, Chromosome 15,

- Chromosomal Triplication, Chromosome Triplication, Chromosome Triploidy Syndrome, Chromosome X, Chromosome XXY, Chronic Acholuric Jaundice, Chronic Adhesive Arachnoiditis, Chronic Adrenocortical Insufficiency, Chronic Cavemosis, Chronic Congenital Aregenerative Anemia, Chronic Dysphagocytosis, Chronic Familial
- 5 Granulomatosis, Chronic Familial Icterus, Chronic Fatigue Immune Dysfunction Syndrome (CFIDS), Chronic Granulomatous Disease, Chronic Guillain-Barre Syndrome, Chronic Idiopathic Jaundice, Chronic Idiopathic Polyneuritis (CIP), Chronic Inflammatory Demyelinating Polyneuropathy, Chronic Inflammatory Demyelinating Polyradiculoneuropathy, Chronic Motor Tic, Chronic Mucocutaneous Candidiasis,
- 10 Chronic Multiple Tics, Chronic Non-Specific Ulcerative Colitis, Chronic Obliterative Cholangitis, Chronic Peptic Ulcer and Esophagitis Syndrome, Chronic Progressive Chorea, Chronic Progressive External Ophthalmoplegia Syndrome, Chronic Progressive External Ophthalmoplegia and myopathy, Chronic Progressive External Ophthalmoplegia with Ragged Red Fibers, Chronic Relapsing Polyneuropathy, Chronic Sarcoidosis, Chronic
- 15 Spasmodic Dysphonia, Chronic Vomiting in Childhood, CHS, Churg-Strauss Syndrome, Cicatricial Pemphigoid, CIP, Cirrhosis Congenital Pigmentary, Cirrhosis, Cistinuria, Citrullinemia, CJD, Classic Schindler Disease, Classic Type Pfeiffer Syndrome, Classical Maple Syrup Urine Disease, Classical Hemophilia, Classical Form Cockayne Syndrome Type I (Type A), Classical Leigh's Disease, Classical Phenylketonuria, Classical X-Linked
- 20 Pelizaeus-Merzbacher Brain Sclerosis, CLE, Cleft Lip/Palate Mucous Cysts Lower Lip PP Digital and Genital Anomalies, Cleft Lip-Palate Blepharophimosis Lagophthalmos and Hypertelorism, Cleft Lip/Palate with Abnormal Thumbs and Microcephaly, Cleft palate-joint contractures-dandy walker malformations, Cleft Palate and Cleft Lip, Cleidocranial Dysplasia w/ Micrognathia, Absent Thumbs, & Distal Aphyalangia, Cleidocranial
- 25 Dysostosis, Cleidocranial Dysplasia, Click murmur syndrome, CLN1, Clonic Spasmodic, Clouston Syndrome, Clubfoot, CMDI, CMM, CMT, CMTC, CMTX, COA Syndrome, Coarctation of the aorta, Coats' Disease, Cobblestone dysplasia, Cochin Jewish Disorder, Cockayne Syndrome, COD-MD Syndrome, COD, Coffin Lowry Syndrome, Coffin Syndrome, Coffin Siris Syndrome, COFS Syndrome, Cogan Corneal Dystrophy, Cogan
- 30 Reese Syndrome, Cohen Syndrome, Cold Agglutinin Disease, Cold Antibody Disease, Cold Antibody Hemolytic Anemia, Colitis Ulcerative, Colitis Gravis, Colitis Ulcerative

- Chronic Non-Specific Ulcerative Colitis, Collodion Baby, Coloboma Heart Defects Atresia of the Choanae Retardation of Growth and Development Genital and Urinary Anomalies and Ear Anomalies, Coloboma, Colonic Neurosis, Color blindness, Colour blindness, Colpocephaly, Columnar-Like Esophagus, Combined Cone-Rod Degeneration, Combined
- 5 Immunodeficiency with Immunoglobulins, Combined Mesoectodermal Dysplasia, Common Variable Hypogammaglobulinemia, Common Variable Immunodeficiency, Common Ventricle, Communicating Hydrocephalus, Complete Absence of Hypoxanthine-Guanine Phosphoribosyltransferase, Complete Atrioventricular Septal Defect, Complement
- 10 Deficiency, Complete Heart Block, Complex Carbohydrate Intolerance, Complex Regional Pain Syndrome, Complex V ATP Synthase Deficiency, Complex I, Complex I NADH dehydrogenase deficiency, Complex II, Complex II Succinate dehydrogenase deficiency, Complex III, Complex III Ubiquinone-cytochrome c oxidoreductase deficiency, Complex IV, Complex IV Cytochrome c oxidase deficiency, Complex IV Deficiency, Complex V,
- 15 Cone-Rod Degeneration, Cone-Rod Degeneration Progressive, Cone Dystrophy, Cone-Rod Dystrophy, Confluent Reticular Papillomatosis, Congenital with low PK Kinetics, Congenital Absence of Abdominal Muscles, Congenital Absence of the Thymus and Parathyroids, Congenital Achromia, Congenital Addison's Disease, Congenital Adrenal Hyperplasia, Congenital Adrenal Hyperplasia, Congenital Afibrinogenemia, Congenital
- 20 Alveolar Hypoventilation, Congenital Anemia of Newborn, Congenital Bilateral Persylvian Syndrome, Congenital Brown Syndrome, Congenital Cardiovascular Defects, Congenital Central Hypoventilation Syndrome, Congenital Cerebral Palsy, Congenital Cervical Synostosis, Congenital Clapsed Thumb with Mental Retardation, Congenital Contractural Arachnodactyly, Congenital Contractures Multiple with Arachnodactyly,
- 25 Congenital Cyanosis, Congenital Defect of the Skull and Scalp, Congenital Dilatation of Intrahepatic Bile Duct, Congenital Dysmyelinating Neuropathy, Congenital Dysphagocytosis, Congenital Dysplastic Angiectasia, Congenital Erythropoietic Porphyria, Congenital Factor XIII Deficiency, Congenital Failure of Autonomic Control of Respiration, Congenital Familial Nonhemolytic Jaundice Type I, Congenital Familial
- 30 Protracted Diarrhea, Congenital Form Cockayne Syndrome Type II (Type B), Congenital Generalized Fibromatosis, Congenital German Measles, Congenital Giant Axonal

Neuropathy, Congenital Heart Block, Congenital Heart Defects, Congenital Hemidysplasia with Ichthyosis Erythroderma and Limb Defects, Congenital Hemolytic Jaundice, Congenital Hemolytic Anemia, Congenital Hepatic Fibrosis, Congenital Hereditary Corneal Dystrophy, Congenital Hereditary Lymphedema, Congenital Hyperchondroplasia, 5 Congenital Hypomyelinating Polyneuropathy, Congenital Hypomyelination Neuropathy, Congenital Hypomyelination, Congenital Hypomyelination (Onion Bulb) Polyneuropathy, Congenital Ichthyosiform Erythroderma, Congenital Keratoconus, Congenital Lactic Acidosis, Congenital Lactose Intolerance, Congenital Lipodystrophy, Congenital Liver Cirrhosis, Congenital Lobar Emphysema, Congenital Localized Emphysema, Congenital 10 Macroglossia, Congenital Medullary Stenosis, Congenital Megacolon, Congenital Melanocytic Nevus, Congenital Mesodermal Dysmorphodystrophy, Congenital Mesodermal Dystrophy, Congenital Microvillus Atrophy, Congenital Multiple Arthrogryposis, Congenital Myotonic Dystrophy, Congenital Neuropathy caused by Hypomyelination, Congenital Pancytopenia, Congenital Pernicious Anemia, Congenital 15 Pernicious Anemia due to Defect of Intrinsic Factor, Congenital Pernicious Anemia due to Defect of Intrinsic Factor, Congenital Pigmentary Cirrhosis, Congenital Porphyria, Congenital Proximal myopathy Associated with Desmin Storage myopathy, Congenital Pulmonary Emphysema, Congenital Pure Red Cell Anemia, Congenital Pure Red Cell Aplasia, Congenital Retinal Blindness, Congenital Retinal Cyst, Congenital Retinitis 20 Pigmentosa, Congenital Retinoschisis, Congenital Rod Disease, Congenital Rubella Syndrome, Congenital Scalp Defects with Distal Limb Reduction Anomalies, Congenital Sensory Neuropathy, Congenital SMA with arthrogryposis, Congenital Spherocytic Anemia, Congenital Spondyloepiphyseal Dysplasia, Congenital Tethered Cervical Spinal Cord Syndrome, Congenital Tyrosinosis, Congenital Varicella Syndrome, Congenital 25 Vascular Cavernous Malformations, Congenital Vascular Veils in the Retina, Congenital Word Blindness, Congenital Wandering Spleen (Pediatric), Congestive Cardio myopathy, Conical Cornea, Conjugated Hyperbilirubinemia, Conjunctivitis, Conjunctivitis Ligneous, Conjunctivo-Urethro-Synovial Syndrome, Conn's Syndrome, Connective Tissue Disease, Conradi Disease, Conradi Hunermann Syndrome, Constitutional Aplastic Anemia, 30 Constitutional Erythroid Hypoplasia, Constitutional Eczema, Constitutional Liver Dysfunction, Constitutional Thrombopathy, Constricting Bands Congenital, Constrictive

Pericarditis with Dwarfism, Continuous Muscle Fiber Activity Syndrome, Contractural Arachnodactyly, Contractures of Feet Muscle Atrophy and Oculomotor Apraxia, Convulsions, Cooley's anemia, Copper Transport Disease, Coproporphyrria Porphyrria Hepatica, Cor Triatriatum, Cor Triatriatum Sinistrum, Cor Triloculare Biatritum, Cor

5 Biloculare, Cori Disease, Cornea Dystrophy, Corneal Amyloidosis, Corneal Clouding-Cutis Laxa-Mental Retardation, Corneal Dystrophy, Cornelia de Lange Syndrome, Coronal Dentine Dysplasia, Coronary Artery Disease, Coronary Heart Disease, Corpus Callosum Agenesis, Cortical-Basal Ganglionic Degeneration, Corticalis Deformaris, Cortico-Basal Ganglionic Degeneration (CBGD), Corticobasal Degeneration, Corticosterone

10 Methloxidase Deficiency Type I, Corticosterone Methyloxidase Deficiency Type II, Cortisol, Costello Syndrome, Cot Death, COVESDEM Syndrome, COX, COX Deficiency, COX Deficiency French-Canadian Type, COX Deficiency Infantile Mitochondrial myopathy de Toni-Fanconi-Debre included, COX Deficiency Type Benign Infantile Mitochondrial Myopathy, CP, CPEO, CPEO with myopathy, CPEO with Ragged-Red

15 Fibers, CPPD Familial Form, CPT Deficiency, CPTD, Cranial Arteritis, Cranial Meningoencephalocele, Cranio-Oro-Digital Syndrome, Craniocarpotarsal dystrophy, Craniocoele, Craniodigital Syndrome-Mental Retardation Scott Type, Craniofacial Dysostosis, Craniofacial Dysostosis-PD Arteriosus-Hypertrichosis-Hypoplasia of Labia, Craniofrontonasal Dysplasia, Craniometaphyseal Dysplasia, Cranioorodigital Syndrome,

20 Cranioorodigital Syndrome Type II, Craniostenosis Crouzon Type, Craniostenosis, Craniosynostosis-Choanal Atresia-Radial Humeral Synostosis, Craniosynostosis-Hypertrichosis-Facial and Other Anomalies, Craniosynostosis Midfacial Hypoplasia and Foot Abnormalities, Craniosynostosis Primary, Craniosynostosis-Radial Aplasia Syndrome, Craniosynostosis with Radial Defects, Cranium Bifidum, CREST Syndrome,

25 Creutzfeldt Jakob Disease, Cri du Chat Syndrome, Crib Death, Crigler Najjar Syndrome Type I, Crohn's Disease, Cronkhite-Canada Syndrome, Cross Syndrome, Cross' Syndrome, Cross-McKusick-Breen Syndrome, Crouzon, Crouzon Syndrome, Crouzon Craniofacial Dysostosis, Cryoglobulinemia Essential Mixed, Cryptophthalmos-Syndactyly Syndrome, Cryptorchidism-Dwarfism-Subnormal Mentality, Crystalline Corneal

30 Dystrophy of Schnyder, CS, CSD, CSID, CSO, CST Syndrome, Curly Hair-Ankyloblephanon-Nail Dysplasia, Curschmann-Batten-Steinert Syndrome, Curth Macklin

- Type Ichthyosis Hystrix, Curth-Macklin Type, Cushing's, Cushing Syndrome, Cushing's
 III, Cutaneous Malignant Melanoma Hereditary, Cutaneous Porphyrias, Cutis Laxa, Cutis
 Laxa-Growth Deficiency Syndrome, Cutis Marmorata Telangiectatica Congenita, CVI,
 CVID, CVS, Cyclic vomiting syndrome, Cystic Disease of the Renal Medulla, Cystic
 5 Hygroma, Cystic Fibrosis, Cystic Lymphangioma, Cystine-Lysine-Arginine-Ornithinuria,
 Cystine Storage Disease, Cystinosis, Cystinuria, Cystinuria with Dibasic Aminoaciduria,
 Cystinuria Type I, Cystinuria Type II, Cystinuria Type III, Cysts of the Renal Medulla
 Congenital, Cytochrome C Oxidase Deficiency, D.C., Dacryosialoadenopathy,
 Dacryosialoadenopathia, Dalpro, Dalton, Daltonism, Danbolt-Cross Syndrome, Dancing
 10 Eyes-Dancing Feet Syndrome, Dandy-Walker Syndrome, Dandy-Walker Cyst, Dandy-
 Walker Deformity, Dandy Walker Malformation, Danish Cardiac Type Amyloidosis (Type
 III), Darier Disease, Davidson's Disease, Davies' Disease, DBA, DBS, DC, DD, De Barsy
 Syndrome, De Barsy-Moens-Diercks Syndrome, de Lange Syndrome, De Morsier
 Syndrome, De Santis Cacchione Syndrome, de Toni-Fanconi Syndrome, Deafness
 15 Congenital and Functional Heart Disease, Deafness-Dwarfism-Retinal Atrophy, Deafness-
 Functional Heart Disease, Deafness Onychodystrophy Osteodystrophy and Mental
 Retardation, Deafness and Pili Torti Bjornstad Type, Deafness Sensorineural with
 Imperforate Anus and Hypoplastic Thumbs, Debrancher Deficiency, Deciduous Skin,
 Defect of Enterocyte Intrinsic Factor Receptor, Defect in Natural Killer Lymphocytes,
 20 Defect of Renal Reabsorption of Carnitine, Deficiency of Glycoprotein Neuraminidase,
 Deficiency of Mitochondrial Respiratory Chain Complex IV, Deficiency of Platelet
 Glycoprotein Ib, Deficiency of Von Willebrand Factor Receptor, Deficiency of Short-
 Chain Acyl-CoA Dehydrogenase (ACADS), Deformity with Mesomelic Dwarfism,
 Degenerative Chorea, Degenerative Lumbar Spinal Stenosis, Degos Disease, Degos-
 25 Kohlmeier Disease, Degos Syndrome, DEH, Dejerine-Roussy Syndrome, Dejerine Sottas
 Disease, Deletion 9p Syndrome Partial, Deletion 11q Syndrome Partial, Deletion 13q
 Syndrome Partial, Delleman-Oorthuys Syndrome, Delleman Syndrome, Dementia with
 Lobar Atrophy and Neuronal Cytoplasmic Inclusions, Demyelinating Disease, DeMyer
 Syndrome, Dentin Dysplasia Coronal, Dentin Dysplasia Radicular, Dentin Dysplasia Type
 30 I, Dentin Dysplasia Type II, Dentinogenesis Imperfecta Brandywine type, Dentinogenesis
 Imperfecta Shields Type, Dentinogenesis Imperfecta Type III, Dento-Oculo-Osseous

- Dysplasia, Dentooculocutaneous Syndrome, Denys-Drash Syndrome, Depakene, DepakeneTM exposure, Depakote, Depakote Sprinkle, Depigmentation-Gingival Fibromatosis-Microphthalmia, Dercum Disease, Dermatitis Atopic, Dermatitis Exfoliativa, Dermatitis Herpetiformis, Dermatitis Multiformis, Dermatochalasia Generalized,
- 5 Dermatology Generalized, Dermatomegaly, Dermatomyositis sine myositis, Dermatomyositis, Dermatosparaxis, Dermatostomatitis Stevens Johnson Type, Desbuquois Syndrome, Desmin Storage myopathy, Desquamation of Newborn, Deuteranomaly, Developmental Reading Disorder, Developmental Gerstmann Syndrome, Devergie Disease, Devic Disease, Devic Syndrome, Dextrocardia- Bronchiectasis and Sinusitis,
- 10 Dextrocardia with Situs Inversus, DGS, DGSX Golabi-Rosen Syndrome Included, DH, DHAP alkyl transferase deficiency, DHBS Deficiency, DHOF, DHPR Deficiency, Diabetes Insipidus, Diabetes Insipidus Diabetes Mellitus Optic Atrophy and Deafness, Diabetes Insipidus Neurohypophyseal, Diabetes Insulin Dependent, Diabetes Mellitus, Diabetes Mellitus Addison's Disease Myxedema, Diabetic Acidosis, Diabetic Bearded
- 15 Woman Syndrome, Diamond-Blackfan Anemia, Diaphragmatic Apnea, Diaphyseal Aclasis, Diastrophic Dwarfism, Diastrophic Dysplasia, Diastrophic Nanism Syndrome, Dicarboxylic Aminoaciduria, Dicarboxylicaciduria Caused by Defect in Beta-Oxidation of Fatty Acids, Dicarboxylicaciduria due to Defect in Beta-Oxidation of Fatty Acids, Dicarboxylicaciduria due to MCADH Deficiency, Dichromasy, Dicker-Opitz, DIDMOAD,
- 20 Diencephalic Syndrome, Diencephalic Syndrome of Childhood, Diencephalic Syndrome of Emaciation, Dienoyl-CoA Reductase Deficiency, Diffuse Cerebral Degeneration in Infancy, Diffuse Degenerative Cerebral Disease, Diffuse Idiopathic Skeletal Hyperostosis, Diffusum-Glycopeptiduria, DiGeorge Syndrome, Digital-Oro-Cranio Syndrome, Digito-Oto-Palatal Syndrome, Digito-Oto-Palatal Syndrome Type I, Digito-Oto-Palatal Syndrome
- 25 Type II, Dihydrobiopterin Synthetase Deficiency, Dihydropteridine Reductase Deficiency, Dihydroxyacetonephosphate synthase, Dilated (Congestive) Cardio myopathy, Dimitri Disease, Diplegia of Cerebral Palsy, Diplo-Y Syndrome, Disaccharidase Deficiency, Disaccharide Intolerance I, Discoid Lupus, Discoid Lupus Erythematosus, DISH, Disorder of Cornification, Disorder of Cornification Type I, Disorder of Cornification 4, Disorder of
- 30 Cornification 6, Disorder of Cornification 8, Disorder of Cornification 9 Netherton's Type, Disorder of Cornification 11 Phytanic Acid Type, Disorder of Cornification 12 (Neutral

Lipid Storage Type), Disorder of Cornification 13, Disorder of Cornification 14, Disorder of Cornification 14 Trichothiodystrophy Type, Disorder of Cornification 15 (Keratitis Deafness Type), Disorder of Cornification 16, Disorder of Cornification 18 Erythrokeratoderma Variabilis Type, Disorder of Cornification 19, Disorder of

5 Cornification 20, Disorder of Cornification 24, Displaced Spleen, Disseminated Lupus Erythematosus, Disseminated Neurodermatitis, Disseminated Sclerosis, Distal 11q Monosomy, Distal 11q- Syndrome, Distal Arthrogryposis Multiplex Congenita Type IIA, Distal Arthrogryposis Multiplex Congenita Type IIA, Distal Arthrogryposis Type IIA, Distal Arthrogryposis Type 2A, Distal Duplication 6q, Distal Duplication 10q, Dup(10q)

10 Syndrome, Distal Duplication 15q, Distal Monosomy 9p, Distal Trisomy 6q, Distal Trisomy 10q Syndrome, Distal Trisomy 11q, Divalproex, DJS, DKC, DLE, DLPIII, DM, DMC Syndrome, DMC Disease, DMD, DNS Hereditary, DOC I, DOC 2, DOC 4, DOC 6 (Harlequin Type), DOC 8 Curth-Macklin Type, DOC 11 Phytanic Acid Type, DOC 12 (Neutral Lipid Storage Type), DOC 13, DOC 14, DOC 14 Trichothiodystrophy Type,

15 DOC 15 (Keratitis Deafness Type), DOC 16, DOC 16 Unilateral Hemidysplasia Type, DOC 18, DOC 19, DOC 20, DOC 24, Dohle's Bodies-Myelopathy, Dolichospondylic Dysplasia, Dolichostenomelia, Dolichostenomelia Syndrome, Dominant Type Kennedy-Caffe Syndrome, Dominant Type Myotonia Congenita, Donahue Syndrome, Donath-Landsteiner Hemolytic Anemia, Donath-Landsteiner Syndrome, DOOR Syndrome,

20 DOORS Syndrome, Dopa-responsive Dystonia (DRD), Dorfman Chanarin Syndrome, Dowling-Meara Syndrome, Down Syndrome, DR Syndrome, Drash Syndrome, DRD, Dreifuss-Emery Type Muscular Dystrophy with Contractures, Dressler Syndrome, Drifting Spleen, Drug-induced Acanthosis Nigricans, Drug-induced Lupus Erythematosus, Drug-related Adrenal Insufficiency, Drummond's Syndrome, Dry Beriberi, Dry Eye, DTD,

25 Duane's Retraction Syndrome, Duane Syndrome, Duane Syndrome Type IA 1B and 1C, Duane Syndrome Type 2A 2B and 2C, Duane Syndrome Type 3A 3B and 3C, Dubin Johnson Syndrome, Dubowitz Syndrome, Duchenne, Duchenne Muscular Dystrophy, Duchenne's Paralysis, Duhring's Disease, Duncan Disease, Duncan's Disease, Duodenal Atresia, Duodenal Stenosis, Duodenitis, Duplication 4p Syndrome, Duplication 6q Partial,

30 Dupuy's Syndrome, Dupuytren's Contracture, Dutch-Kennedy Syndrome, Dwarfism, Dwarfism Campomelic, Dwarfism Cortical Thickening of the Tubular Bones & Transient

- Hypocalcemia, Dwarfism Levi's Type, Dwarfism Metatropic, Dwarfism-Onychodysplasia, Dwarfism-Pericarditis, Dwarfism with Renal Atrophy and Deafness, Dwarfism with Rickets, DWM, Dyggve Melchior Clausen Syndrome, Dysautonomia Familial, Dysbetalipoproteinemia Familial, Dyschondrodysplasia with Hemangiomas,
- 5 Dyschondrosteosis, Dyschromatosis Universalis Hereditaria, Dysencephalia Splanchnocystica, Dyskeratosis Congenita, Dyskeratosis Congenita Autosomal Recessive, Dyskeratosis Congenita Scoggins Type, Dyskeratosis Congenita Syndrome, Dyskeratosis Follicularis Vegetans, Dyslexia, Dysmyelogenic Leukodystrophy, Dysmyelogenic Leukodystrophy-Megalobare, Dysphonia Spastica, Dysplasia Epiphysialis Punctata,
- 10 Dysplasia Epiphyseal Hemimelica, Dysplasia of Nails With Hypodontia, Dysplasia Cleidocranial, Dysplasia Fibrous, Dysplasia Gigantism SyndromeX-Linked, Dysplasia Osteodental, Dysplastic Nevus Syndrome, Dysplastic Nevus Type, Dyssynergia Cerebellaris Myoclonica, Dyssynergia Esophagus, Dystonia, Dystopia Canthorum, Dystrophia Adiposogenitalis, Dystrophia Endothelialis Cornea, Dystrophia Mesodermalis,
- 15 Dystrophic Epidermolysis Bullosa, Dystrophy, Asphyxiating Thoracic, Dystrophy Myotonic, E-D Syndrome, Eagle-Barrett Syndrome, Eales Retinopathy, Eales Disease, Ear Anomalies-Contractures-Dysplasia of Bone with Kyphoscoliosis, Ear Patella Short Stature Syndrome, Early Constraint Defects, Early Hypercalcemia Syndrome with Elfin Facie, Early-onset Dystonia, Eaton Lambert Syndrome, EB, Ebstein's anomaly, EBV
- 20 Susceptibility (EBVS), EBVS, ECD, ECPSG, Ectodermal Dysplasias, Ectodermal Dysplasia Anhidrotic with Cleft Lip and Cleft Palate, Ectodermal Dysplasia-Exocrine Pancreatic Insufficiency, Ectodermal Dysplasia Rapp-Hodgkin type, Ectodermal and Mesodermal Dysplasia Congenital, Ectodermal and Mesodermal Dysplasia with Osseous Involvement, Ectodermosis Erosiva Pluriorificialis, Ectopia Lentis, Ectopia Vesicae,
- 25 Ectopic ACTH Syndrome, Ectopic Adrenocorticotropic Hormone Syndrome, Ectopic Anus, Ectrodactilia of the Hand, Ectrodactyly, Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome, Ectrodactyly Ectodermal Dysplasias Clefting Syndrome, Ectrodactyly Ectodermal Dysplasia Cleft Lip/Cleft Palate, Eczema, Eczema-Thrombocytopenia-Immunodeficiency Syndrome, EDA, EDMD, EDS, EDS Arterial-Ecchymotic Type, EDS
- 30 Arthrochalasia, EDS Classic Severe Form, EDS Dysfibronectinemic, EDS Gravis Type, EDS Hypermobility, EDS Kyphoscoliotic, EDS Kyphoscoliosis, EDS Mitis Type, EDS

- Ocular-Scoliotic, EDS Progeroid, EDS Periodontosis, EDS Vascular, EEC Syndrome, EFE, EHBA, EHK, Ehlers Danlos Syndrome, Ehlers-Danlos syndrome, Ehlers Danlos IX, Eisenmenger Complex, Eisenmenger's complex, Eisenmenger Disease, Eisenmenger Reaction, Eisenmenger Syndrome, Ekblom Syndrome, Ekman-Lobstein Disease,
- 5 Ektrodactyly of the Hand, EKV, Elastin fiber disorders, Elastorrhexis Generalized, Elastosis Dystrophica Syndrome, Elective Mutism (obsolete), Elective Mutism, Electrocardiogram (ECG or EKG), Electron Transfer Flavoprotein (ETF) Dehydrogenase Deficiency: (GAI & MADD), Electrophysiologic study (EPS), Elephant Nails From Birth, Elephantiasis Congenita Angiomatosa, Hemangiectatic Hypertrophy, Elfin Facies with
- 10 Hypercalcemia, Ellis-van Creveld Syndrome, Ellis Van Creveld Syndrome, Embryoma Kidney, Embryonal Adenomyosarcoma Kidney, Embryonal Carcinosarcoma Kidney, Embryonal Mixed Tumor Kidney, EMC, Emery Dreyfus Muscular Dystrophy, Emery-Dreifuss Muscular Dystrophy, Emery-Dreifuss Syndrome, EMF, EMG Syndrome, Empty Sella Syndrome, Encephalitis Periaxialis Diffusa, Encephalitis Periaxialis Concentrica,
- 15 Encephalocele, Encephalofacial Angiomatosis, Encephalopathy, Encephalotrigeminal Angiomatosis, Enchondromatosis with Multiple Cavernous Hemangiomas, Endemic Polyneuritis, Endocardial Cushion Defect, Endocardial Cushion Defects, Endocardial Dysplasia, Endocardial Fibroelastosis (EFE), Endogenous Hypertriglyceridemia, Endolymphatic Hydrops, Endometrial Growths, Endometriosis, Endomyocardial Fibrosis,
- 20 Endothelial Corneal Dystrophy Congenital, Endothelial Epithelial Corneal Dystrophy, Endothelium, Engelmann Disease, Enlarged Tongue, Enterocolitis, Enterocyte Cobalamin Malabsorption, Eosinophilia Syndrome, Eosinophilic Cellulitis, Eosinophilic Fasciitis, Eosinophilic Granuloma, Eosinophilic Syndrome, Epidermal Nevus Syndrome, Epidermolysis Bullosa, Epidermolysis Bullosa Acquisita, Epidermolysis Bullosa
- 25 Hereditaria, Epidermolysis Bullosa Letalis, Epidermolysis Hereditaria Tarda, Epidermolytic Hyperkeratosis, Epidermolytic Hyperkeratosis (Bullous CIE), Epilepsia Procursiva, Epilepsy, Epinephrine, Epiphyseal Changes and High Myopia, Epiphyseal Osteochondroma Benign, Epiphysealis Hemimelica Dysplasia, Episodic-Abnormal Eye Movement, Epithelial Basement Membrane Corneal Dystrophy, Epithelial Corneal
- 30 Dystrophy of Meesmann Juvenile, Epitheliomatosis Multiplex with Nevus, Epithelium, Epival, EPS, Epstein-Barr Virus-Induced Lymphoproliferative Disease in Males, Erb-

- Goldflam syndrome, Erdheim Chester Disease, Erythema Multiforme Exudativum, Erythema Polymorphe Stevens Johnson Type, Erythroblastophthisis, Erythroblastosis Fetalis, Erythroblastosis Neonatorum, Erythroblastotic Anemia of Childhood, Erythrocyte Phosphoglycerate Kinase Deficiency, Erythrogenesis Imperfecta, Erythrokeratoderma
- 5 Progressiva Symmetrica, Erythrokeratoderma Progressiva Symmetrica Ichthyosis, Erythrokeratoderma Variabilis, Erythrokeratoderma Variabilis Type, Erythrokeratolysis Hiemalis, Erythropoietic Porphyrias, Erythropoietic Porphyria, Escobar Syndrome, Esophageal Atresia, Esophageal Aperistalsis, Esophagitis-Peptic Ulcer, Esophagus Atresia and/or Tracheoesophageal Fistula, Essential Familial Hyperlipemia, Essential Fructosuria,
- 10 Essential Hematuria, Essential Hemorrhagic Thrombocythemia, Essential Mixed Cryoglobulinemia, Essential Moschowitz Disease, Essential Thrombocythemia, Essential Thrombocytopenia, Essential Thrombocytosis, Essential Tremor, Esterase Inhibitor Deficiency, Estren-Dameshek variant of Fanconi Anemia, Estrogen-related Cholestasis, ET, ETF, Ethylmalonic Adipicaciduria, Eulenburg Disease, pc, EVCS, Exaggerated Startle
- 15 Reaction, Exencephaly, Exogenous Hypertriglyceridemia, Exomphalos-Macroglossia-Gigantism Syndrom, Exophthalmic Goiter, Expanded Rubella Syndrome, Exstrophy of the Bladder, EXT, External Chondromatosis Syndrome, Extrahépatie Biliary Atresia, Extramedullary Plasmacytoma, Exudative Retinitis, Eye Retraction Syndrome, FA1, FAA, Fabry Disease, FAC, FACB, FACD, FACE, FACF, FACG, FACH, Facial Nerve Palsy,
- 20 Facial Paralysis, Facial Ectodermal Dysplasias, Facial Ectodermal Dysplasia, Facio-Scapulo-Humeral Dystrophy, Facio-Auriculo-Vertebral Spectrum, Facio-cardio-cutaneous syndrome, Facio-Fronto-Nasal Dysplasia, Faciocutaneoskeletal Syndrome, Faciodigitogenital syndrome, Faciogenital dysplasia, Faciogenitopopliteal Syndrome, Faciopalatoosseous Syndrome, Faciopalatoosseous Syndrome Type II,
- 25 Facioscapulohumeral muscular dystrophy, Factitious Hypoglycemia, Factor VIII Deficiency, Factor IX Deficiency, Factor XI Deficiency, Factor XII deficiency, Factor XIII Deficiency, Fahr Disease, Fahr's Disease, Failure of Secretion Gastric Intrinsic Factor, Fairbank Disease, Fallot's Tetralogy, Familial Acrogeria, Familial Acromicria, Familial Adenomatous Colon Polyposis, Familial Adenomatous Polyposis with Extraintestinal
- 30 Manifestations, Familial Alobar Holoprosencephaly, Familial Alpha-Lipoprotein Deficiency, Familial Amyotrophic Chorea with Acanthocytosis, Familial Arrhythmic

- Myoclonus, Familial Articular Chondrocalcinosis, Familial Atypical Mole-Malignant Melanoma Syndrome, Familial Broad Beta Disease, Familial Calcium Gout, Familial Calcium Pyrophosphate Arthropathy, Familial Chronic Obstructive Lung Disease, Familial Continuous Skin Peeling, Familial Cutaneous Amyloidosis, Familial Dysproteinemia,
- 5 Familial Emphysema, Familial Enteropathy Microvillus, Familial Foveal Retinoschisis, Familial Hibernation Syndrome, Familial High Cholesterol, Familial Hemochromatosis, Familial High Blood Cholesterol, Familial High-Density Lipoprotein Deficiency, Familial High Serum Cholesterol, Familial Hyperlipidemia, Familial Hypoproteinemia with Lymphangietatic Enteropathy, Familial Jaundice, Familial Juvenile Nephronophthisis-
- 10 Associated Ocular Anomaly, Familial Lichen Amyloidosis (Type IX), Familial Lumbar Stenosis, Familial Lymphedema Praecox, Familial Mediterranean Fever, Familial Multiple Polyposis, Familial Nuchal Bleb, Familial Paroxysmal Polyserositis, Familial Polyposis Coli, Familial Primary Pulmonary Hypertension, Familial Renal Glycosuria, Familial Splenic Anemia, Familial Startle Disease, Familial Visceral Amyloidosis (Type VIII),
- 15 FAMMM, FANCA, FANCB, FANCC, FANCD, FANCE, Fanconi Panmyelopathy, Fanconi Pancytopenia, Fanconi II, Fanconi's Anemia, Fanconi's Anemia Type I, Fanconi's Anemia Complementation Group, Fanconi's Anemia Complementation Group^{CKA}, Fanconi's Anemia Complementation Group B, Fanconi's Anemia Complementation Group C, Fanconi's Anemia Complementation Group D, Fanconi's Anemia Complementation
- 20 Group E, Fanconi's Anemia Complementation Group G, Fanconi's Anemia Complementation Group H, Fanconi's Anemia Estren-Dameshek Variant, FANF, FANG, FANH, FAP, FAPG, Farber's Disease, Farber's Lipogranulomatosis, FAS, Fasting Hypoglycemia, Fat-Induced Hyperlipemia, Fatal Granulomatous Disease of Childhood, Fatty Oxidation Disorders, Fatty Liver with Encephalopathy, FAV, FCH, FCMD, FCS
- 25 Syndrome, FD, FDH, Febrile Mucocutaneous Syndrome Stevens Johnson Type, Febrile Neutrophilic Dermatitis Acute, Febrile Seizures, Feinberg's syndrome, Feissinger-Leroy-Reiter Syndrome, Female Pseudo-Turner Syndrome, Femoral Dysgenesis Bilateral-Robin Anomaly, Femoral Dysgenesis Bilateral, Femoral Facial Syndrome, Femoral Hypoplasia-Unusual Facies Syndrome, Fetal Alcohol Syndrome, Fetal Anti-Convulsant Syndrome,
- 30 Fetal Cystic Hygroma, Fetal Effects of Alcohol, Fetal Effects of Chickenpox, Fetal Effects of Thalidomide, Fetal Effects of Varicella Zoster Virus, Fetal Endomyocardial Fibrosis,

Fetal Face Syndrome, Fetal Iritis Syndrome, Fetal Transfusion Syndrome, Fetal Valproate Syndrome, Fetal Valproic Acid Exposure Syndrome, Fetal Varicella Infection, Fetal Varicella Zoster Syndrome, FFDD Type II, FG Syndrome, FGDY, FHS, Fibrin Stabilizing Factor Deficiency, Fibrinase Deficiency, Fibrinoid Degeneration of Astrocytes, Fibrinoid

5 Leukodystrophy, Fibrinogase Deficiency, Fibroblastoma Perineural, Fibrocystic Disease of Pancreas, Fibrodysplasia Ossificans Progressiva, Fibroelastic Endocarditis, Fibromyalgia, Fibromyalgia-Fibromyositis, Fibromyositis, Fibrosing Cholangitis, Fibrositis, Fibrous Ankylosis of Multiple Joints, Fibrous Cavernositis, Fibrous Dysplasia, Fibrous Plaques of the Penis, Fibrous Sclerosis of the Penis, Fickler-Winkler Type, Fiedler

10 Disease, Fifth Digit Syndrome, Filippi Syndrome, Finnish Type Amyloidosis (Type V), First Degree Congenital Heart Block, First and Second Branchial Arch Syndrome, Fischer's Syndrome, Fish Odor Syndrome, Fissured Tongue, Flat Adenoma Syndrome, Flatau-Schilder Disease, Flavin Containing Monooxygenase 2, Floating Beta Disease, Floating-Harbor Syndrome, Floating Spleen, Floppy Infant Syndrome, Floppy Valve

15 Syndrome, Fluent aphasia, FMD, FMF, FMO Adult Liver Form, FMO2, FND, Focal Dermal Dysplasia Syndrome, Focal Dermal Hypoplasia, Focal Dermato-Phalangeal Dysplasia, Focal Dystonia, Focal Epilepsy, Focal Facial Dermal Dysplasia Type II, Focal Neuromyotonia, FODH, Folling Syndrome, Fong Disease, FOP, Forbes Disease, Forbes-Albright Syndrome, Forestier's Disease, Forsius-Eriksson Syndrome (X-Linked),

20 Fothergill Disease, Fountain Syndrome, Foveal Dystrophy Progressive, FPO Syndrome Type II, FPO, Fraccaro Type Achondrogenesis (Type IB), Fragile X syndrome, Franceschetti-Zwahlen-Klein Syndrome, Francois Dyscephaly Syndrome, Francois-Neetens Speckled Dystrophy, Flecked Corneal Dystrophy, Fraser Syndrome, FRAXA, FRDA, Fredrickson Type I Hyperlipoproteinemia, Freeman-Sheldon Syndrome, Freire-Maia

25 Syndrome, Frey's Syndrome, Friedreich's Ataxia, Friedreich's Disease, Friedreich's Tabes, FRNS, Froelich's Syndrome, Frommel-Chiari Syndrome, Frommel-Chiari Syndrome Lactation-Uterus Atrophy, Frontodigital Syndrome, Frontofacionasal Dysostosis, Frontofacionasal Dysplasia, Frontonasal Dysplasia, Frontonasal Dysplasia with Coronal Craniosynostosis, Fructose-1-Phosphate Aldolase Deficiency, Fructosemia,

30 Fructosuria, Fryns Syndrome, FSH, FSHD, FSS, Fuchs Dystrophy, Fucosidosis Type 1, Fucosidosis Type 2, Fucosidosis Type 3, Fukuhara Syndrome, Fukuyama Disease,

- Fukuyama Type Muscular Dystrophy, Fumarylacetoacetase deficiency, Furrowed Tongue, G Syndrome, G6PD Deficiency, G6PD, GA I, GA IIB, GA IIA, GA II, GAI & MADD, Galactorrhea-Amenorrhea Syndrome Nonpuerperal, Galactorrhea-Amenorrhea without Pregnancy, Galactosamine-6-Sulfatase Deficiency, Galactose-1-Phosphate Uridyl
- 5 Transferase Deficiency, Galactosemia, GALB Deficiency, Galloway-Mowat Syndrome, Galloway Syndrome, GALT Deficiency, Gammaglobulin Deficiency, GAN, Ganglioside Neuraminidase Deficiency, Ganglioside Sialidase Deficiency, Gangliosidosis GM1 Type 1, Gangliosidosis GM2 Type 2, Gangliosidosis Beta Hexosaminidase B Deficiency, Gardner Syndrome, Gargoylism, Garies-Mason Syndrome, Gasser Syndrome, Gastric
- 10 Intrinsic Factor Failure of Secretion, Enterocyte Cobalamin, Gastrinoma, Gastritis, Gastroesophageal Laceration-Hemorrhage, Gastrointestinal Polyposis and Ectodermal Changes, Gastroschisis, Gaucher Disease, Gaucher-Schlagenhauser, Gayet-Wernicke Syndrome, GBS, GCA, GCM Syndrome, GCPS, Gee-Herter Disease, Gee-Thaysen Disease, Gehrig's Disease, Gelineau's Syndrome, Genee-Wiedemann Syndrome,
- 15 Generalized Dystonia, Generalized Familial Neuromyotonia, Generalized Fibromatosis, Generalized Flexion Epilepsy, Generalized Glycogenosis, Generalized Hyperhidrosis, Generalized Lipofuscinosis, Generalized Myasthenia Gravis, Generalized Myotonia, Generalized Sporadic Neuromyotonia, Genetic Disorders, Genital Defects, Genital and Urinary Tract Defects, Gerstmann Syndrome, Gerstmann Tetrad, GHBP, GHD, GHR,
- 20 Giant Axonal Disease, Giant Axonal Neuropathy, Giant Benign Lymphoma, Giant Cell Glioblastoma Astrocytoma, Giant Cell Arteritis, Giant Cell Disease of the Liver, Giant Cell Hepatitis, Giant Cell of Newborns Cirrhosis, Giant Cyst of the Retina, Giant Lymph Node Hyperplasia, Giant Platelet Syndrome Hereditary, Giant Tongue, gic Macular Dystrophy, Gilbert's Disease, Gilbert Syndrome, Gilbert-Dreyfus Syndrome, Gilbert-
- 25 Lereboullet Syndrome, Gilford Syndrome, Gilles de la Tourette's syndrome, Gillespie Syndrome, Gingival Fibromatosis-Abnormal Fingers Nails Nose Ear Splenomegaly, GLA Deficiency, GLA, GLB1, Glioma Retina, Global aphasia, Globoid Leukodystrophy, Glossoptosis Micrognathia and Cleft Palate, Glucocerebrosidase deficiency, Glucocerebrosidosis, Glucose-6-Phosphate Dehydrogenase Deficiency, Glucose-6-
- 30 Phosphate Transport Defect, Glucose-6-Phosphate Translocase Deficiency, Glucose-G-Phosphatase Deficiency, Glucose-Galactose Malabsorption, Glucosyl Ceramide Lipidosis,

- Glutaric Aciduria I, Glutaric Acidemia I, Glutaric Acidemia II, Glutaric Aciduria II, Glutaric Aciduria Type II, Glutaric Aciduria Type III, Glutaricacidemia I, Glutaricacidemia II, Glutaricaciduria I, Glutaricaciduria II, Glutaricaciduria Type IIA, Glutaricaciduria Type IIB, Glutaryl-CoA Dehydrogenase Deficiency, Glutaurate-Aspartate
- 5 Transport Defect, Gluten-Sensitive Enteropathy, Glycogen Disease of Muscle Type VII, Glycogen Storage Disease I, Glycogen Storage Disease III, Glycogen Storage Disease IV, Glycogen Storage Disease Type V, Glycogen Storage Disease VI, Glycogen Storage Disease VII, Glycogen Storage Disease VIII, Glycogen Storage Disease Type II, Glycogen Storage Disease-Type II, Glycogenosis, Glycogenosis Type I, Glycogenosis Type IA,
- 10 Glycogenosis Type IB, Glycogenosis Type II, Glycogenosis Type II, Glycogenosis Type III, Glycogenosis Type IV, Glycogenosis Type V, Glycogenosis Type VI, Glycogenosis Type VII, Glycogenosis Type VIII, Glycolic Aciduria, Glycolipid Lipidosis, GM2 Gangliosidosis Type 1, GM2 Gangliosidosis Type 1, GNPTA, Goitrous Autoimmune Thyroiditis, Goldenhar Syndrome, Goldenhar-Gorlin Syndrome, Goldscheider's Disease,
- 15 Goltz Syndrome, Goltz-Gorlin Syndrome, Gonadal Dysgenesis 45 X, Gonadal Dysgenesis XO, Goniodysgenesis-Hypodontia, Goodman Syndrome, Goodman, Goodpasture Syndrome, Gordon Syndrome, Gorlin's Syndrome, Gorlin-Chaudhry-Moss Syndrome, Gottron Erythrokeratoderma Congenitalis Progressiva Symmetrica, Gottron's Syndrome, Gougerot-Carteaud Syndrome, Graftversus Host Disease, Grand Mal Epilepsy, Granular
- 20 Type Corneal Dystrophy, Granulomatous Arteritis, Granulomatous Colitis, Granulomatous Dermatitis with Eosinophilia, Granulomatous Ileitis, Graves Disease, Graves' Hyperthyroidism, Graves' Disease, Greig Cephalopolysyndactyly Syndrome, Groenouw Type I Corneal Dystrophy, Groenouw Type II Corneal Dystrophy, Gronblad-Strandberg Syndrome, Grotton Syndrome, Growth Hormone Receptor Deficiency, Growth Hormone
- 25 Binding Protein Deficiency, Growth Hormone Deficiency, Growth-Mental Deficiency Syndrome of Myhre, Growth Retardation-Rieger Anomaly, GRS, Gruber Syndrome, GS, GSD6, GSD8, GTS, Guanosine Triphosphate-Cyclohydrolase Deficiency, Guanosine Triphosphate-Cyclohydrolase Deficiency, Guenther Porphyria, Guerin-Stem Syndrome, Guillain-Barré, Guillain-Barre Syndrome, Gunther Disease, H Disease, H. Gottron's
- 30 Syndrome, Habit Spasms, HAE, Hageman Factor Deficiency, Hageman factor, Haim-Munk Syndrome, Hajdu-Cheney Syndrome, Hajdu Cheney, HAL Deficiency, Hall-

- Pallister Syndrome, Hallermann-Streiff-Francois syndrome, Hallermann-Streiff Syndrome, Hallervorden-Spatz Disease, Hallervorden-Spatz Syndrome, Hallopeau-Siemens Disease, Hallux Duplication Postaxial Polydactyly and Absence of Corpus Callosum, Halushi-
 Behcet's Syndrome, Hamartoma of the Lymphatics, Hand-Schueller-Christian Syndrome,
 5 HANE, Hanhart Syndrome, Happy Puppet Syndrome, Harada Syndrome, HARD +/-E
 Syndrome, HARD Syndrome, Hare Lip, Harlequin Fetus, Harlequin Type DOC 6,
 Harlequin Type Ichthyosis, Harley Syndrome, Harrington Syndrome, Hart Syndrome,
 Hartnup Disease, Hartnup Disorder, Hartnup Syndrome, Hashimoto's Disease, Hashimoto-
 Pritzker Syndrome, Hashimoto's Syndrome, Hashimoto's Thyroiditis, Hashimoto-Pritzker
 10 Syndrome, Hay Well's Syndrome, Hay-Wells Syndrome of Ectodermal Dysplasia,
 HCMM, HCP, HCTD, HD, Heart-Hand Syndrome (Holt-Oram Type), Heart Disease,
 Hecht Syndrome, HED, Heerferdt-Waldenstrom and Lofgren's Syndromes, Hegglin's
 Disease, Heinrichsbauer Syndrome, Hemangiomas, Hemangioma Familial, Hemangioma-
 Thrombocytopenia Syndrome, Hemangiomatosis Chondrodystrophica, Hemangiomatous
 15 Branchial Clefts-Lip Pseudocleft Syndrome, Hemifacial Microsomia,
 Hemimegalencephaly, Hemiparesis of Cerebral Palsy, Hemiplegia of Cerebral Palsy,
 Hemisection of the Spinal Cord, Hemochromatosis, Hemochromatosis Syndrome,
 Hemodialysis-Related Amyloidosis, Hemoglobin Lepore Syndromes, Hemolytic Anemia
 of Newborn, Hemolytic Cold Antibody Anemia, Hemolytic Disease of Newborn,
 20 Hemolytic-Uremic Syndrome, Hemophilia, Hemophilia A, Hemophilia B, Hemophilia B
 Factor IX, Hemophilia C, Hemorrhagic Dystrophic Thrombocytopenia, Hemorrhagica
 Aleukia, Hemosiderosis, Hepatic Fructokinase Deficiency, Hepatic Phosphorylase Kinase
 Deficiency, Hepatic Porphyria, Hepatic Porphyrias, Hepatic Veno-Occlusive Diseases,
 Hepato-Renal Syndrome, Hepatolenticular Degeneration, Hepatophosphorylase
 25 Deficiency, Hepatorenal Glycogenosis, Hepatorenal Syndrome, Hepatorenal Tyrosinemia,
 Hereditary Acromelalgia, Hereditary Alkaptonuria, Hereditary Amyloidosis, Hereditary
 Angioedema, Hereditary Areflexic Dystasia, Heredopathia Atactica Polyneuritiformis,
 Hereditary Ataxia, Hereditary Ataxia Friedrich's Type, Hereditary Benign Acanthosis
 Nigricans, Hereditary Cerebellar Ataxia, Hereditary Chorea, Hereditary Chronic
 30 Progressive Chorea, Hereditary Connective Tissue Disorders, Hereditary Coproporphyruria,
 Hereditary Coproporphyruria Porphyria, Hereditary Cutaneous Malignant Melanoma,

- Hereditary Deafness-Retinitis Pigmentosa, Heritable Disorder of Zinc Deficiency, Hereditary DNS, Hereditary Dystopic Lipidosis, Hereditary Emphysema, Hereditary Fructose Intolerance, Hereditary Hemorrhagic Telangiectasia, Hereditary Hemorrhagic Telangiectasia Type I, Hereditary Hemorrhagic Telangiectasia Type II, Hereditary Hemorrhagic Telangiectasia Type III, Hereditary Hyperuricemia and Choreaethetosis Syndrome, Hereditary Leptocytosis Major, Hereditary Leptocytosis Minor, Hereditary Lymphedema, Hereditary Lymphedema Tarda, Hereditary Lymphedema Type I, Hereditary Lymphedema Type II, Hereditary Motor Sensory Neuropathy, Hereditary Motor Sensory Neuropathy I, Hereditary Motor Sensory Neuropathy Type III, Hereditary Nephritis, Hereditary Nephritis and Nerve Deafness, Hereditary Nephropathic Amyloidosis, Hereditary Nephropathy and Deafness, Hereditary Nonpolyposis Colorectal Cancer, Hereditary Nonpolyposis Colorectal Carcinoma, Hereditary Nonspherocytic Hemolytic Anemia, Hereditary Onychoosteodysplasia, Hereditary Optic Neuroretinopathy, Hereditary Polyposis Coli, Hereditary Sensory and Autonomic Neuropathy Type I, Hereditary Sensory and Autonomic Neuropathy Type II, Hereditary Sensory and Autonomic Neuropathy Type III, Hereditary Sensory Motor Neuropathy, Hereditary Sensory Neuropathy type I, Hereditary Sensory Neuropathy Type I, Hereditary Sensory Neuropathy Type II, Hereditary Sensory Neuropathy Type III, Hereditary Sensory Radicular Neuropathy Type I, Hereditary Sensory Radicular Neuropathy Type I, Hereditary Sensory Radicular Neuropathy Type II, Hereditary Site Specific Cancer, Hereditary Spherocytic Hemolytic Anemia, Hereditary Spherocytosis, Hereditary Tyrosinemia Type 1, Heritable Connective Tissue Disorders, Herlitz Syndrome, Hermans-Herzberg Phakomatosis, Hermansky-Pudlak Syndrome, Hermaphroditism, Herpes Zoster, Herpes Iris Stevens-Johnson Type, Hers Disease, Heterozygous Beta Thalassemia, Hexoaminidase Alpha-Subunit Deficiency (Variant B), Hexoaminidase Alpha-Subunit Deficiency (Variant B), HFA, HFM, HGPS, HH, HHHO, HHRH, HHT, Hiatal Hernia-Microcephaly-Nephrosis Galloway Type, Hidradenitis Suppurativa, Hidrosadenitis Axillaris, Hidrosadenitis Suppurativa, Hidrotic Ectodermal Dysplasias, HIE Syndrome, High Imperforate Anus, High Potassium, High Scapula, HIM, Hirschsprung's Disease, Hirschsprung's Disease Acquired, Hirschsprung Disease Polydactyly of Ulnar & Big Toe and VSD, Hirschsprung Disease with Type D Brachydactyly, Hirsutism, HIS Deficiency,

- Histidine Ammonia-Lyase (HAL) Deficiency, Histidase Deficiency, Histidinemia, Histiocytosis, Histiocytosis X, HLHS, HLP Type II, HMG, HMI, HMSN I, HNHA, HOCM, Hodgkin Disease, Hodgkin's Disease, Hodgkin's Lymphoma, Hollaender-Simons Disease, Holmes-Adie Syndrome, Holocarboxylase Synthetase Deficiency,
- 5 Holoprosencephaly, Holoprosencephaly Malformation Complex, Holoprosencephaly Sequence, Holt-Oram Syndrome, Holt-Oram Type Heart-Hand Syndrome, Homocystinemia, Homocystinuria, Homogentisic Acid Oxidase Deficiency, Homogentisic Aciduria, Homozygous Alpha-1-Antitrypsin Deficiency, HOOD, Horner Syndrome, Horton's disease, HOS, HOS1, Houston-Harris Type Achromodrogenesis (Type IA), HPS,
- 10 HRS, HS, HSAN Type I, HSAN Type II, HSAN-III, HSMN, HSMN Type III, HSN I, HSN-III, Huebner-Herter Disease, Hunner's Patch, Hunner's Ulcer, Hunter Syndrome, Hunter-Thompson Type Acromesomelic Dysplasia, Huntington's Chorea, Huntington's Disease, Hurler Disease, Hurler Syndrome, Hurler-Scheie Syndrome, HUS, Hutchinson-Gilford Progeria Syndrome, Hutchinson-Gilford Syndrome, Hutchinson-Weber-Peutz
- 15 Syndrome, Hutterite Syndrome Bowen-Conradi Type, Hyaline Panneuropathy, Hydranencephaly, Hydrocephalus, Hydrocephalus Agyria and Retinal Dysplasia, Hydrocephalus Internal Dandy-Walker Type, Hydrocephalus Noncommunicating Dandy-Walker Type, Hydrocephaly, Hydronephrosis With Peculiar Facial Expression, Hydroxylase Deficiency, Hygroma Colli, Hyper-IgE Syndrome, Hyper-IgM Syndrome,
- 20 Hyperaldosteronism, Hyperaldosteronism With Hypokalemic Alkalosis, Hyperaldosteronism Without Hypertension, Hyperammonemia, Hyperammonemia Due to Carbamylphosphate Synthetase Deficiency, Hyperammonemia Due to Ornithine Transcarbamylase Deficiency, Hyperammonemia Type II, Hyper-Beta Carnosinemia, Hyperbilirubinemia I, Hyperbilirubinemia II, Hypercalcemia Familial with
- 25 Nephrocalcinosis and Indicanuria, Hypercalcemia-Supravalvar Aortic Stenosis, Hypercalciuric Rickets, Hypercapnic acidosis, Hypercatabolic Protein-Losing Enteropathy, Hyperchloremic acidosis, Hypercholesterolemia, Hypercholesterolemia Type IV, Hyperchylomicronemia, Hypercystinuria, Hyperekplexia, Hyperextensible joints, Hyperglobulinemic Purpura, Hyperglycinemia with Ketoacidosis and Lactic Acidosis
- 30 Propionic Type, Hyperglycinemia Nonketotic, Hypergonadotropic Hypogonadism, Hyperimmunoglobulin E Syndrome, Hyperimmunoglobulin E-Recurrent Infection

Syndrome, Hyperimmunoglobulinemia E-Staphylococcal, Hyperkalemia, Hyperkinetic Syndrome, Hyperlipemic Retinitis, Hyperlipidemia I, Hyperlipidemia IV, Hyperlipoproteinemia Type I, Hyperlipoproteinemia Type III, Hyperlipoproteinemia Type IV, Hyperoxaluria, Hyperphalangy-Clinodactyly of Index Finger with Pierre Robin Syndrome, Hyperphenylalanemia, Hyperplastic Epidermolysis Bullosa, Hyperpnea, 5 Hyperpotassemia, Hyperprebeta-Lipoproteinemia, Hyperprolinemia Type I, Hyperprolinemia Type II, Hypersplenism, Hypertelorism with Esophageal Abnormalities and Hypospadias, Hypertelorism-Hypospadias Syndrome, Hypertrophic Cardio myopathy, Hypertrophic Interstitial Neuropathy, Hypertrophic Interstitial Neuritis, Hypertrophic 10 Interstitial Radiculoneuropathy, Hypertrophic Neuropathy of Refsum, Hypertrophic Obstructive Cardio myopathy, Hyperuricemia Choreoathetosis Self-mutilation Syndrome, Hyperuricemia-Oligophrenia, Hypervalinemia, Hypocalcified (Hypomineralized) Type, Hypochondrogenesis, Hypochondroplasia, Hypogammaglobulinemia, Hypogammaglobulinemia Transient of Infancy, Hypogenital Dystrophy with Diabetic 15 Tendency, Hypoglossia-Hypodactylia Syndrome, Hypoglycemia, Exogenous Hypoglycemia, Hypoglycemia with Macroglossia, Hypoglycosylation Syndrome Type 1a, Hypoglycosylation Syndrome Type 1a, Hypogonadism with Anosmia, Hypogonadotropic Hypogonadism and Anosmia, Hypohidrotic Ectodermal Dysplasia, Hypohidrotic Ectodermal Dysplasia Autosomal Dominant type, Hypohidrotic Ectodermal Dysplasias 20 Autorecessive, Hypokalemia, Hypokalemic Alkalosis with Hypercalciuria, Hypokalemic Syndrome, Hypolactasia, Hypomaturation Type (Snow-Capped Teeth), Hypomelanosis of Ito, Hypomelia-Hypotrichosis-Facial Hemangioma Syndrome, Hypomyelination Neuropathy, Hypoparathyroidism, Hypophosphatasia, Hypophosphatemic Rickets with Hypercalcemia, Hypopigmentation, Hypopigmented macular lesion, Hypoplasia of the 25 Depressor Anguli Oris Muscle with Cardiac Defects, Hypoplastic Anemia, Hypoplastic Congenital Anemia, Hypoplastic Chondrodystrophy, Hypoplastic Enamel-Onycholysis-Hypohidrosis, Hypoplastic (Hypoplastic-Expilastic) Type, Hypoplastic Left Heart Syndrome, Hypoplastic-Triphalangeal Thumbs, Hypopotassemia Syndrome, Hypospadias-Dysphagia Syndrome, Hyposmia, Hypothalamic Hamartoblastoma Hypopituitarism 30 Imperforate Anus Polydactyly, Hypothalamic Infantilism-Obesity, Hypothyroidism, Hypotonia-Hypomentia-Hypogonadism-Obesity Syndrome, Hypoxanthine-Guanine

- Phosphoribosyltransferase Defect (Complete Absence of), I-Cell Disease, Iatrogenic Hypoglycemia, IBGC, IBIDS Syndrome, IBM, IBS, IC, I-Cell Disease, ICD, ICE Syndrome Cogan-Reese Type, Icelandic Type Amyloidosis (Type VI), I-Cell Disease, Ichthyosiform Erythroderma Corneal Involvement and Deafness, Ichthyosiform
- 5 Erythroderma Hair Abnormality Growth and Men, Ichthyosiform Erythroderma with Leukocyte Vacuolation, Ichthyosis, Ichthyosis Congenita, Ichthyosis Congenital with Trichothiodystrophy, Ichthyosis Hystrix, Ichthyosis Hystrix Gravior, Ichthyosis Linearis Circumflexa, Ichthyosis Simplex, Ichthyosis Tay Syndrome, Ichthyosis Vulgaris, Ichthyotic Neutral Lipid Storage Disease, Icteric Leptospirosis, Icterohemorrhagic
 - 10 Leptospirosis, Icterus (Chronic Familial), Icterus Gravis Neonatorum, Icterus Intermittens Juvenalis, Idiopathic Alveolar Hypoventilation, Idiopathic Amyloidosis, Idiopathic Arteritis of Takayasu, Idiopathic Basal Ganglia Calcification (IBGC), Idiopathic Brachial Plexus Neuropathy, Idiopathic Cervical Dystonia, Idiopathic Dilatation of the Pulmonary Artery, Idiopathic Facial Palsy, Idiopathic Familial Hyperlipemia, Idiopathic Hypertrophic
 - 15 Subaortic Stenosis, Idiopathic Hypoproteinemia, Idiopathic Immunoglobulin Deficiency, Idiopathic Neonatal Hepatitis, Idiopathic Non-Specific Ulcerative Colitis, Idiopathic Peripheral Periphlebitis, Idiopathic Pulmonary Fibrosis, Idiopathic Refractory Sideroblastic Anemia, Idiopathic Renal Hematuria, Idiopathic Steatorrhea, Idiopathic Thrombocythemia, Idiopathic Thrombocytopenic Purpura, Idiopathic Thrombocytopenia
 - 20 Purpura (ITP), IDPA, IgA Nephropathy, IHSS, Ileitis, Ileocolitis, Illinois Type Amyloidosis, ILS, IM, IMD2, IMD5, Immune Defect due to Absence of Thymus, Immune Hemolytic Anemia Paroxysmal Cold, Immunodeficiency with Ataxia Telangiectasia, Immunodeficiency Cellular with Abnormal Immunoglobulin Synthesis, Immunodeficiency Common Variable Unclassifiable, Immunodeficiency with Hyper-IgM, Immunodeficiency
 - 25 with Leukopenia, Immunodeficiency-2, Immunodeficiency-5 (IMD5), Immunoglobulin Deficiency, Imperforate Anus, Imperforate Anus with Hand Foot and Ear Anomalies, Imperforate Nasolacrimal Duct and Premature Aging Syndrome, Impotent Neutrophil Syndrome, Inability To Open Mouth Completely And Short Finger-Flexor, INAD, Inborn Error of Urea Synthesis Arginase Type, Inborn Error of Urea Synthesis Arginino Succinic
 - 30 Type, Inborn Errors of Urea Synthesis Carbamyl Phosphate Type, Inborn Error of Urea Synthesis Citrullinemia Type, Inborn Errors of Urea Synthesis Glutamate Synthetase Type,

- 68 -

INCL, Inclusion body myositis, Incomplete Atrioventricular Septal Defect, Incomplete Testicular Feminization, Incontinentia Pigmenti, Incontinenti Pigmenti Achromians, Index Finger Anomaly with Pierre Robin Syndrome, Indiana Type Amyloidosis (Type II), Indolent systemic mastocytosis, Infantile Acquired Aphasia, Infantile Autosomal

5 Recessive Polycystic Kidney Disease, Infantile Beriberi, Infantile Cerebral Ganglioside, Infantile Cerebral Paralysis, Infantile Cystinosis, Infantile Epileptic, Infantile Fanconi Syndrome with Cystinosis, Infantile Finnish Type Neuronal Ceroid Lipofuscinosis, Infantile Gaucher Disease, Infantile Hypoglycemia, Infantile Hypophosphatasia, Infantile

10 Lobar Emphysema, Infantile Myoclonic Encephalopathy, Infantile Myoclonic Encephalopathy and Polymyoclonia, Infantile Myofibromatosis, Infantile Necrotizing Encephalopathy, Infantile Neuronal Ceroid Lipofuscinosis, Infantile Neuroaxonal Dystrophy, Infantile Onset Schindler Disease, Infantile Phytanic Acid Storage Disease, Infantile Refsum Disease (IRD), Infantile Sipoidosis GM-2 Gangliosidosis (Type S), Infantile Sleep Apnea, Infantile Spasms, Infantile Spinal Muscular Atrophy (all types),

15 Infantile Spinal Muscular Atrophy ALS, Infantile Spinal Muscular Atrophy Type I, Infantile Type Neuronal Ceroid Lipofuscinosis, Infectious Jaundice, Inflammatory Breast Cancer, Inflammatory Linear Nevus Sebaceous Syndrome, Iniencephaly, Insulin Resistant Acanthosis Nigrans, Insulin Lipodystrophy, Insulin dependent Diabetes, Intention Myoclonus, Intermediate Cystinosis, Intermediate Maple Syrup Urine Disease, Intermittent

20 Ataxia with Pyruvate Dehydrogenase Deficiency, Intermittent Maple Syrup Urine Disease, Internal Hydrocephalus, Interstitial Cystitis, Interstitial Deletion of 4q Included, Intestinal Lipodystrophy, Intestinal Lipophagic Granulomatosis, Intestinal Lymphangiectasia, Intestinal Polyposis I, Intestinal Polyposis II, Intestinal Polyposis III, Intestinal Polyposis-Cutaneous Pigmentation Syndrome, Intestinal Pseudoobstruction with External

25 Ophthalmoplegia, Intracranial Neoplasm, Intracranial Tumors, Intracranial Vascular Malformations, Intrauterine Dwarfism, Intrauterine Synechiae, Inverted Smile And Occult Neuropathic Bladder, Iowa Type Amyloidosis (Type IV), IP, IPA, Iridocorneal Endothelial Syndrome, Iridocorneal Endothelial (ICE) Syndrome Cogan-Resse Type, Iridogoniodysgenesis With Somatic Anomalies, Iris Atrophy with Corneal Edema and

30 Glaucoma, Iris Nevus Syndrome, Iron Overload Anemia, Iron Overload Disease, Irritable Bowel Syndrome, Irritable Colon Syndrome, Isaacs Syndrome, Isaacs-Merten Syndrome,

- Ischemic Cardio myopathy, Isolated Lissencephaly Sequence, Isoleucine 33 Amyloidosis, Isovaleric Acid CoA Dehydrogenase Deficiency, Isovaleric Acidaemia, Isovalericacidemia, Isovaleryl CoA Carboxylase Deficiency, ITO Hypomelanosis, ITO, ITP, IVA, Ivemark Syndrome, Iwanoff Cysts, Jackknife Convulsion, Jackson-Weiss
- 5 Craniosynostosis, Jackson-Weiss Syndrome, Jacksonian Epilepsy, Jacobsen Syndrome, Jadassohn-Lewandowsky Syndrome, Jaffe-Lichenstein Disease, Jakob's Disease, Jakob-Creutzfeldt Disease, Janeway I, Janeway Dysgammaglobulinemia, Jansen Metaphyseal Dysostosis, Jansen Type Metaphyseal Chondrodysplasia, Jarcho-Levin Syndrome, Jaw-Winking, JBS, JDMS, Jegher's Syndrome, Jejunal Atresia, Jejunitis, Jejunoileitis, Jervell
- 10 and Lange-Nielsen Syndrome, Jeune Syndrome, JMS, Job Syndrome, Job-Buckley Syndrome, Johanson-Blizzard Syndrome, John Dalton, Johnson-Stevens Disease, Jonston's Alopecia, Joseph's Disease, Joseph's Disease Type I, Joseph's Disease Type II, Joseph's Disease Type III, Joubert Syndrome, Joubert-Bolthausen Syndrome, JRA, Juberg
- 15 Hayward Syndrome, Juberg-Marsidi Syndrome, Juberg-Marsidi Mental Retardation Syndrome, Jumping Frenchmen, Jumping Frenchmen of Maine, Juvenile Arthritis, Juvenile Autosomal Recessive Polycystic Kidney Disease, Juvenile Cystinosis, Juvenile (Childhood) Dermatomyositis (JDMS), Juvenile Diabetes, Juvenile Gaucher Disease, Juvenile Gout Choreoathetosis and Mental Retardation Syndrome, Juvenile Intestinal
- 20 Malabsorption of Vit B12, Juvenile Intestinal Malabsorption of Vitamin B12, Juvenile Macular Degeneration, Juvenile Pernicious Anemia, Juvenile Retinoschisis, Juvenile Rheumatoid Arthritis, Juvenile Spinal Muscular Atrophy Included, Juvenile Spinal Muscular Atrophy ALS Included, Juvenile Spinal Muscular Atrophy Type III, Juxta-Articular Adiposis Dolorosa, Juxtaglomerular Hyperplasia, Kabuki Make-Up Syndrome, Kahler Disease, Kallmann Syndrome, Kanner Syndrome, Kanzaki Disease, Kaposi Disease
- 25 (not Kaposi Sarcoma), Kappa Light Chain Deficiency, Karsch-Neugebauer Syndrome, Kartagener Syndrome-Chronic Sinobronchial Disease and Dextrocardia, Kartagener Triad, Kasabach-Merritt Syndrome, Kast Syndrome, Kawasaki Disease, Kawasaki Syndrome, KBG Syndrome, KD, Kearns-Sayre Disease, Kearns-Sayre Syndrome, Kennedy Disease, Kennedy Syndrome, Kennedy Type Spinal and Bulbar Muscular Atrophy, Kennedy-
- 30 Stefanis Disease, Kenny Disease, Kenny Syndrome, Kenny Type Tubular Stenosis, Kenny-Caffe Syndrome, Kera. Palmoplant. Con. Pes Planus Ony. Periodon. Arach.,

- Keratitis Ichthyosis Deafness Syndrome, Keratoconus, Keratoconus Posticus Circumscriptus, Keratolysis, Keratolysis Exfoliativa Congenita, Keratolytic Winter Erythema, Keratomalacia, Keratosis Follicularis, Keratosis Follicularis Spinulosa Decalvans, Keratosis Follicularis Spinulosa Decalvans Ichthyosis, Keratosis Nigricans,
- 5 Keratosis Palmoplantaris with Periodontopathia and Onychogryposis, Keratosis Palmoplantaris Congenital Pes Planus Onychogryposis Periodontosis Arachnodactyly, Keratosis Palmoplantaris Congenital, Pes Planus, Onychogryphosis, Periodontosis, Arachnodactyly, Acroosteolysis, Keratosis Rubra Figurata, Keratosis Seborrheica, Ketoacid Decarboxylase Deficiency, Ketoaciduria, Ketotic Glycinemia, KFS, KID
- 10 Syndrome, Kidney Agenesis, Kidneys Cystic-Retinal Aplasia Joubert Syndrome, Killian Syndrome, Killian/Teschler-Nicola Syndrome, Kiloh-Nevin syndrome III, Kinky Hair Disease, Kinsbourne Syndrome, Kleeblattschadel Deformity, Kleine-Levin Syndrome, Kleine-Levin Hibernation Syndrome, Klinefelter, Klippel-Feil Syndrome, Klippel-Feil Syndrome Type I, Klippel-Feil Syndrome Type II, Klippel-Feil Syndrome Type III,
- 15 Klippel Trenaunay Syndrome, Klippel-Trenaunay-Weber Syndrome, Kluver-Bucy Syndrome, KMS, Kniest Dysplasia, Kniest Syndrome, Kobner's Disease, Koebberling-Dunnigan Syndrome, Kohlmeier-Degos Disease, Kok Disease, Korsakoff Psychosis, Korsakoff's Syndrome, Krabbe's Disease Included, Krabbe's Leukodystrophy, Kramer Syndrome, KSS, KTS, KTW Syndrome, Kufs Disease, Kugelberg-Welander Disease,
- 20 Kugelberg-Welander Syndrome, Kussmaul-Landry Paralysis, KWS, L-3-Hydroxy-Acyl-CoA Dehydrogenase (LCHAD) Deficiency, Laband Syndrome, Labhart-Willi Syndrome, Labyrinthine Syndrome, Labyrinthine Hydrops, Lacrimo-Auriculo-Dento-Digital Syndrome, Lactase Isolated Intolerance, Lactase Deficiency, Lactation-Uterus Atrophy, Lactic Acidosis Leber Hereditary Optic Neuropathy, Lactic and Pyruvate Acidemia with
- 25 Carbohydrate Sensitivity, Lactic and Pyruvate Acidemia with Episodic Ataxia and Weakness, Lactic and Pyruvate, Lactic acidosis, Lactose Intolerance of Adulthood, Lactose Intolerance, Lactose Intolerance of Childhood, LADD Syndrome, LADD, Lafora Disease Included, Lafora Body Disease, Laki-Lorand Factor Deficiency, LAM, Lambert Type Ichthyosis, Lambert-Eaton Syndrome, Lambert-Eaton Myasthenic Syndrome,
- 30 Lamellar Recessive Ichthyosis, Lamellar Ichthyosis, Lancereaux-Mathieu-Weil Spirochetosis, Landau-Kleffner Syndrome, Landouzy Dejerine Muscular Dystrophy,

Landry Ascending Paralysis, Langer-Salidino Type Achondrogensis (Type II), Langer
 Giedion Syndrome, Langerhans-Cell Granulomatosis, Langerhans-Cell Histiocytosis
 (LCH), Large Atrial and Ventricular Defect, Laron Dwarfism, Laron Type Pituitary
 Dwarfism, Larsen Syndrome, Laryngeal Dystonia, Latah (Observed in Malaysia), Late
 5 Infantile Neuroaxonal Dystrophy, Late Infantile Neuroaxonal Dystrophy, Late Onset
 Cockayne Syndrome Type III (Type C), Late-Onset Dystonia, Late-Onset Immunoglobulin
 Deficiency, Late Onset Pelizaeus-Merzbacher Brain Sclerosis, Lattice Corneal Dystrophy,
 Lattice Dystrophy, Launois-Bensaude, Launois-Cleret Syndrome, Laurence Syndrome,
 Laurence-Moon Syndrome, Laurence-Moon/Bardet-Biedl, Lawrence-Seip Syndrome,
 10 LCA, LCAD Deficiency, LCAD, LCAD, LCADH Deficiency, LCH, LCHAD, LCPD, Le
 Jeune Syndrome, Leband Syndrome, Leber's Amaurosis, Leber's Congenital
 Amaurosis, Congenital Absence of the Rods and Cones, Leber's Congenital Tapetoretinal
 Degeneration, Leber's Congenital Tapetoretinal Dysplasia, Leber's Disease, Leber's Optic
 Atrophy, Leber's Optic Neuropathy, Left Ventricular Fibrosis, Leg Ulcer, Legg-Calve-
 15 Perthes Disease, Leigh's Disease, Leigh's Syndrome, Leigh's Syndrome (Subacute
 Necrotizing Encephalomyelopathy), Leigh Necrotizing Encephalopathy, Lennox-Gastaut
 Syndrome, Lentigio-Polypose-Digestive Syndrome, Lenz Dysmorphogenetic Syndrome,
 Lenz Dysplasia, Lenz Microphthalmia Syndrome, Lenz Syndrome, LEOPARD Syndrome,
 Leprechaunism, Leptomeningeal Angiomatosis, Leptospiral Jaundice, Leri-Weill Disease,
 20 Leri-Weil Dyschondrosteosis, Leri-Weil Syndrome, Lermoyez Syndrome, Leroy Disease,
 Lesch Nyhan Syndrome, Lethal Infantile Cardio myopathy, Lethal Neonatal Dwarfism,
 Lethal Osteochondrodysplasia, Letterer-Siwe Disease, Leukocytic Anomaly Albinism,
 Leukocytic Inclusions with Platelet Abnormality, Leukodystrophy, Leukodystrophy with
 Rosenthal Fibers, Leukoencephalitis Periaxialis Concentric, Levine-Critchley Syndrome,
 25 Levulosuria, Levy-Hollister Syndrome, LGMD, LGS, LHON, LIC, Lichen Ruber
 Acuminatus, Lichen Acuminatus, Lichen Amyloidosis, Lichen Planus, Lichen Psoriasis,
 Lignac-Debre-Fanconi Syndrome, Lignac-Fanconi Syndrome, Ligneous Conjunctivitis,
 Limb-Girdle Muscular Dystrophy, Limb Malformations-Dento-Digital Syndrome, Limit
 Dextrinosis, Linear Nevroid Hypermelanosis, Linear Nevus Sebaceous Syndrome, Linear
 30 Scleroderma, Linear Sebaceous Nevus Sequence, Linear Sebaceous Nevus Syndrome,
 Lingua Fissurata, Lingua Plicata, Lingua Scrotalis, Linguofacial Dyskinesia, Lip

- Pseudocleft-hemangiomatous Branchial Cyst Syndrome, Lipid Granulomatosis, Lipid Histiocytosis, Lipid Kerasin Type, Lipid Storage Disease, Lipid-Storage myopathy Associated with SCAD Deficiency, Lipidosis Ganglioside Infantile, Lipoatrophic Diabetes Mellitus, Lipodystrophy, Lipoid Corneal Dystrophy, Lipoid Hyperplasia-Male
- 5 Pseudohermaphroditism, Lipomatosis of Pancreas Congenital, Lipomucopolysaccharidosis Type I, Lipomyelomeningocele, Lipoprotein Lipase Deficiency Familial, LIS, LIS1, Lissencephaly 1, Lissencephaly Type I, Lissencephaly variants with agenesis of the corpus callosum cerebellar hypoplasia or other anomalies, Little Disease, Liver Phosphorylase Deficiency, LKS, LM Syndrome, Lobar Atrophy, Lobar Atrophy of the Brain, Lobar
- 10 Holoprosencephaly, Lobar Tension Emphysema in Infancy, Lobstein Disease (Type I), Lobster Claw Deformity, Localized Epidermolysis Bullosa, Localized Lipodystrophy, Localized Neuritis of the Shoulder Girdle, Loeffler's Disease, Loeffler Endomyocardial Fibrosis with Eosinophilia, Loeffler Fibroplastic Parietal Endocarditis, Loken Syndrome, Loken-Senior Syndrome, Long-Chain 3-hydroxyacyl-CoA Dehydrogenase (LCHAD),
- 15 Long Chain Acyl CoA Dehydrogenase Deficiency, Long-Chain Acyl-CoA Dehydrogenase (ACADL), Long-Chain Acyl-CoA Dehydrogenase Deficiency, Long QT Syndrome without Deafness, Lou Gehrig's Disease, Lou Gehrig's Disease Included, Louis-Bar Syndrome, Low Blood Sugar, Low-Density Beta Lipoprotein Deficiency, Low Imperforate Anus, Low Potassium Syndrome, Lowe syndrome, Lowe's Syndrome, Lowe-Bickel
- 20 Syndrome, Lowe-Terry-MacLachlan Syndrome, LS, LTD, Lubs Syndrome, Luft Disease, Lumbar Canal Stenosis, Lumbar Spinal Stenosis, Lumbosacral Spinal Stenosis, Lundborg-Unverricht Disease, Lundborg-Unverricht Disease Included, Lupus, Lupus Erythematosus, Luschka-Magendie Foramina Atresia, Lyell Syndrome, Lyelles Syndrome, Lymphadenoid Goiter, Lymphangiectatic Protein-Losing Enteropathy, Lymphangioma, Lymphangioma
- 25 Lymphangioma, Lymphatic Malformations, Lynch Syndromes, Lynch Syndrome I, Lynch Syndrome II, Lysosomal Alpha-N-Acetylgalactosaminidase Deficiency Schindler Type, Lysosomal Glycoaminoacid Storage Disease-Angiokeratoma Corporis Diffusum, Lysosomal Glucosidase Deficiency, MAA, Machado Disease, Machado-Joseph Disease, Macrencephaly, Macrocephaly,
- 30 Macrocephaly Hemihypertrophy, Macrocephaly with Multiple Lipomas and Hemangiomas, Macrocephaly with Pseudopapilledema and Multiple Hemangiomas,

- Macroglobulinemia, Macroglossia, Macroglossia-Omphalocele-Visceromegaly Syndrome, Macrostomia Ablepharon Syndrome, Macrothrombocytopenia Familial Bernard-Soulier Type, Macula Lutea degeneration, Macular Amyloidosis, Macular Degeneration, Macular Degeneration Disciform, Macular Degeneration Senile, Macular Dystrophy, Macular Type
- 5 Corneal Dystrophy, MAD, Madelung's Disease, Maffucci Syndrome, Major Epilepsy, Malabsorption, Malabsorption-Ectodermal Dysplasia-Nasal Alar Hypoplasia, Maladie de Roger, Maladie de Tics, Male Malformation of Limbs and Kidneys, Male Turner Syndrome, Malignant Acanthosis, Malignant Acanthosis Nigricans, Malignant Astrocytoma, Malignant Atrophic Papulosis, Malignant Fever, Malignant
- 10 Hyperphenylalaninemia, Malignant Hyperpyrexia, Malignant Hyperthermia, Malignant Melanoma, Malignant Tumors of the Central Nervous System, Mallory-Weiss Laceration, Mallory-Weiss Tear, Mallory-Weiss Syndrome, Mammary Paget's Disease, Mandibular Ameloblastoma, Mandibulofacial Dysostosis, Manic Depression Illness Disease, Mannosidosis, Map-Dot-Fingerprint Type Corneal Dystrophy, Maple Syrup Urine Disease,
- 15 Marble Bones, Marchiafava-Micheli Syndrome, Marcus Gunn Jaw-Winking Syndrome, Marcus Gunn Phenomenon, Marcus Gunn Ptosis with jaw-winking, Marcus Gunn Syndrome, Marcus Gunn (Jaw-Winking) Syndrome, Marcus Gunn Ptosis (with jaw-winking), Marden-Walker Syndrome, Marden-Walker Type Connective Tissue Disorder, Marfan's Abiotrophy, Marfan-Achard syndrome, Marfan Syndrome, Marfan's Syndrome
- 20 I, Marfan's Variant, Marfanoid Hypermobility Syndrome, Marginal Corneal Dystrophy, Marie's Ataxia, Marie Disease, Marie-Sainton Disease, Marie Strumpell Disease, Marie-Strumpell Spondylitis, Marinesco-Sjogren Syndrome, Marinesco-Sjogren-Gorland Syndrome, Marker X Syndrome, Maroteaux Lamy Syndrome, Maroteaux Type Acromesomelic Dysplasia, Marshall's Ectodermal Dysplasias With Ocular and Hearing
- 25 Defects, Marshall-Smith Syndrome, Marshall Syndrome, Marshall Type Deafness-Myopia-Cataract-Saddle Nose, Martin-Albright Syndrome, Martin-Bell Syndrome, Martorell Syndrome, MASA Syndrome, Massive Myoclonia, Mast Cell Leukemia, Mastocytosis, Mastocytosis With an Associated Hematologic Disorder, Maumenee Corneal Dystrophy, Maxillary Ameloblastoma, Maxillofacial Dysostosis, Maxillonasal
- 30 Dysplasia, Maxillonasal Dysplasia Binder Type, Maxillopalpebral Synkinesis, May-Hegglin Anomaly, MCAD Deficiency, MCAD, McArdle Disease, McCune-Albright,

MCD, McKusick Type Metaphyseal Chondrodysplasia, MCR, MCTD, Meckel Syndrome, Meckel-Gruber Syndrome, Median Cleft Face Syndrome, Mediterranean Anemia, Medium-Chain Acyl-CoA dehydrogenase (ACADM), Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency, Medium-Chain Acyl-CoA Dehydrogenase
5 Deficiency, Medullary Cystic Disease, Medullary Sponge Kidney, MEF, Megaesophagus, Megalencephaly, Megalencephaly with Hyaline Inclusion, Megalencephaly with Hyaline Panneuropathy, Megaloblastic Anemia, Megaloblastic Anemia of Pregnancy, Megalocornea-Mental Retardation Syndrome, Meier-Gorlin Syndrome, Meige's Lymphedema, Meige's Syndrome, Melanodermic Leukodystrophy, Melanoplakia-
10 Intestinal Polyposis, Melanoplakia-Intestinal Polyposis, MELAS Syndrome, MELAS, Melkersson Syndrome, Melnick-Fraser Syndrome, Melnick-Needles Osteodysplasty, Melnick-Needles Syndrome, Membranous Lipodystrophy, Mendes Da Costa Syndrome, Meniere Disease, Ménière's Disease, Meningeal Capillary Angiomas, Menkes Disease, Menke's Syndrome I, Mental Retardation Aphasia Shuffling Gait Adducted Thumbs
15 (MASA), Mental Retardation-Deafness-Skeletal Abnormalities-Coarse Face with Full Lips, Mental Retardation with Hypoplastic 5th Fingernails and Toenails, Mental Retardation with Osteocartilaginous Abnormalities, Mental Retardation-X-linked with Growth Delay-Deafness-Microgenitalism, Menzel Type OPCA, Mermaid Syndrome, MERRF, MERRF Syndrome, Merten-Singleton Syndrome, MES, Mesangial IGA
20 Nephropathy, Mesenteric Lipodystrophy, Mesiodens-Cataract Syndrome, Mesodermal Dysmorphodystrophy, Mesomelic Dwarfism-Madelung Deformity, Metabolic Acidosis, Metachromatic Leukodystrophy, Metatarsus Varus, Metatropic Dwarfism Syndrome, Metatropic Dysplasia, Metatropic Dysplasia I, Metatropic Dysplasia II, Methylmalonic Acidemia, Methylmalonic Aciduria, Meulengracht's Disease, MFD1, MG, MH, MHA,
25 Micrencephaly, Microcephalic Primordial Dwarfism I, Microcephaly, Microcephaly-Hiatal Hernia-Nephrosis Galloway Type, Microcephaly-Hiatal Hernia-Nephrotic Syndrome, Microcystic Corneal Dystrophy, Microcythemia, Microlissencephaly, Microphthalmia, Microphthalmia or Anophthalmos with Associated Anomalies, Micropolygyria With Muscular Dystrophy, Microtia Absent Patellae Micrognathia Syndrome, Microvillus
30 Inclusion Disease, MID, Midsystolic-click-late systolic murmur syndrome, Miescher's Type I Syndrome, Mikulicz Syndrome, Mikulicz-Radecki Syndrome, Mikulicz-Sjogren

- Syndrome, Mild Autosomal Recessive, Mild Intermediate Maple Syrup Urine Disease, Mild Maple Syrup Urine Disease, Miller Syndrome, Miller-Dieker Syndrome, Miller-Fisher Syndrome, Milroy Disease, Minkowski-Chauffard Syndrome, Minor Epilepsy, Minot-Von Willebrand Disease, Mirror-Image Dextrocardia, Mitochondrial Beta-
5 Oxidation Disorders, Mitochondrial and Cytosolic, Mitochondrial Cytopathy, Mitochondrial Cytopathy, Kearns-Sayre Type, Mitochondrial Encephalopathy, Mitochondrial Encephalo myopathy Lactic Acidosis and Strokelike Episodes, Mitochondrial myopathy, Mitochondrial myopathy Encephalopathy Lactic Acidosis Stroke-Like Episode, Mitochondrial PEPCK Deficiency, Mitral-valve prolapse, Mixed
10 Apnea, Mixed Connective Tissue Disease, Mixed Hepatic Porphyria, Mixed Non-Fluent Aphasia, Mixed Sleep Apnea, Mixed Tonic and Clonic Torticollis, MJD, MKS, ML I, ML II, ML III, ML IV, ML Disorder Type I, ML Disorder Type II, ML Disorder Type III, ML Disorder Type IV, MLNS, MMR Syndrome, MND, MNGIE, MNS, Mobitz I, Mobitz II, Mobius Syndrome, Moebius Syndrome, Moersch-Woltmann Syndrome, Mohr Syndrome,
15 Monilethrix, Monomodal Visual Amnesia, Mononeuritis Multiplex, Mononeuritis Peripheral, Mononeuropathy Peripheral, Monosomy 3p2, Monosomy 9p Partial, Monosomy 11q Partial, Monosomy 13q Partial, Monosomy 18q Syndrome, Monosomy 22, Monostotic Fibrous Dysplasia, Morgagni-Turner-Albright Syndrome, Morphea, Morquio Disease, Morquio Syndrome, Morquio Syndrome A, Morquio Syndrome B, Morquio-
20 Brailsford Syndrome, Morvan Disease, Mosaic Tetrasomy 9p, Motor Neuron Disease, Motor Neuron Syndrome, Motor Neurone Disease, Motoneuron Disease, Motoneurone Disease, Motor System Disease (Focal and Slow), Moya-moya Disease, Moyamoya Disease, MPS, MPS I, MPS I H, MPS I H/S Hurler/Scheie Syndrome, MPS I S Scheie Syndrome, MPS II, MPS IIA, MPS IIB, MPS II-AR Autosomal Recessive Hunter
25 Syndrome, MPS II-XR, MPS II-XR Severe Autosomal Recessive, MPS III, MPS III A B C and D Sanfilippo A, MPS IV, MPS IV A and B Morquio A, MPS V, MPS VI, MPS VI Severe Intermediate Mild Maroteaux-Lamy, MPS VII, MPS VII Sly Syndrome, MPS VIII, MPS Disorder, MPS Disorder I, MPS Disorder II, MPS Disorder III, MPS Disorder VI, MPS Disorder Type VII, MRS, MS, MSA, MSD, MSL, MSS, MSUD, MSUD, MSUD
30 Type Ib, MSUD Type II, Mucocutaneous Lymph Node Syndrome, Mucopolipidosis I, Mucopolipidosis II, Mucopolipidosis III, Mucopolipidosis IV, Mucopolysaccharidosis,

- Mucopolysaccharidosis I-H, Mucopolysaccharidosis I-S, Mucopolysaccharidosis II, Mucopolysaccharidosis III, Mucopolysaccharidosis IV, Mucopolysaccharidosis VI, Mucopolysaccharidosis VII, Mucopolysaccharidosis Type I, Mucopolysaccharidosis Type II, Mucopolysaccharidosis Type III, Mucopolysaccharidosis Type VII, Mucosis,
- 5 Mucosulfatidosis, Mucous Colitis, Mucoviscidosis, Mulibrey Dwarfism, Mulibrey Nanism Syndrome, Mullerian Duct Aplasia-Renal Aplasia-Cervicothoracic Somite Dysplasia, Mullerian Duct-Renal-Cervicothoracic-Upper Limb Defects, Mullerian Duct and Renal Agenesis with Upper Limb and Rib Anomalies, Mullerian-Renal-Cervicothoracic Somite Abnormalities, Multi-Infarct Dementia Binswanger's Type, Multicentric Castleman's
- 10 Disease, Multifocal Eosinophilic Granuloma, Multiple Acyl-CoA Dehydrogenase Deficiency, Multiple Acyl-CoA Dehydrogenase Deficiency / Glutaric Aciduria Type II, Multiple Angiomas and Endochondromas, Multiple Carboxylase Deficiency, Multiple Cartilaginous Enchondroses, Multiple Cartilaginous Exostoses, Multiple Enchondromatosis, Multiple Endocrine Deficiency Syndrome Type II, Multiple
- 15 Epiphyseal Dysplasia, Multiple Exostoses, Multiple Exostoses Syndrome, Multiple Familial Polyposis, Multiple Lentigines Syndrome, Multiple Myeloma, Multiple Neuritis of the Shoulder Girdle, Multiple Osteochondromatosis, Multiple Peripheral Neuritis, Multiple Polyposis of the Colon, Multiple Pterygium Syndrome, Multiple Sclerosis, Multiple Sulfatase Deficiency, Multiple Symmetric Lipomatosis, Multiple System
- 20 Atrophy, Multisynostotic Osteodysgenesis, Multisynostotic Osteodysgenesis with Long Bone Fractures, Mulvihill-Smith Syndrome, MURCS Association, Murk Jansen Type Metaphyseal Chondrodysplasia, Muscle Carnitine Deficiency, Muscle Core Disease, Muscle Phosphofructokinase Deficiency, Muscular Central Core Disease, Muscular Dystrophy, Muscular Dystrophy Classic X-linked Recessive, Muscular Dystrophy
- 25 Congenital With Central Nervous System Involvement, Muscular Dystrophy Congenital Progressive with Mental Retardation, Muscular Dystrophy Facioscapulohumeral, Muscular Rheumatism, Muscular Rigidity - Progressive Spasm, Musculoskeletal Pain Syndrome, Mutilating Acropathy, Mutism, mvp, MVP, MWS, Myasthenia Gravis, Myasthenia Gravis Pseudoparalytica, Myasthenic Syndrome of Lambert-Eaton, Myelinoclastic Diffuse
- 30 Sclerosis, Myelomatosis, Myhre Syndrome, Myoclonic Astatic Petit Mal Epilepsy, Myoclonic Dystonia, Myoclonic Encephalopathy of Infants, Myoclonic Epilepsy,

- Myoclonic Epilepsy Hartung Type, Myoclonus Epilepsy Associated with Ragged Red
Fibers, Myoclonic Epilepsy and Ragged-Red Fiber Disease, Myoclonic Progressive
Familial Epilepsy, Myoclonic Progressive Familial Epilepsy, Myoclonic Seizure,
Myoclonus, Myoclonus Epilepsy, Myoencephalopathy Ragged-Red Fiber Disease,
5 Myofibromatosis, Myofibromatosis Congenital, Myogenic Facio-Scapulo-Peroneal
Syndrome, Myoneurogastrointestinal Disorder and Encephalopathy, Myopathic
Arthrogryposis Multiplex Congenita, Myopathic Carnitine Deficiency, Myopathy Central
Fibrillar, myopathy Congenital Nonprogressive, myopathy Congenital Nonprogressive
with Central Axis, myopathy with Deficiency of Carnitine Palmitoyltransferase,
10 myopathy-Marinesco-Sjogren Syndrome, myopathy-Metabolic Carnitine
Palmitoyltransferase Deficiency, myopathy Mitochondrial-Encephalopathy-Lactic
Acidosis-Stroke, myopathy with Sarcoplasmic Bodies and Intermediate Filaments,
Myophosphorylase Deficiency, Myositis Ossificans Progressiva, Myotonia Atrophica,
Myotonia Congenita, Myotonia Congenita Intermittens, Myotonic Dystrophy, Myotonic
15 myopathy Dwarfism Chondrodystrophy Ocular and Facial Anomalies, Myotubular
myopathy, Myotubular myopathy X-linked, Myproic Acid, Myriachit (Observed in
Siberia), Myxedema, N-Acetylglucosamine-1-Phosphotransferase Deficiency, N-Acetyl
Glutamate Synthetase Deficiency, NADH-CoQ reductase deficiency, Naegeli Ectodermal
Dysplasias, Nager Syndrome, Nager Acrofacial Dysostosis Syndrome, Nager Syndrome,
20 NAGS Deficiency, Nail Dystrophy-Deafness Syndrome, Nail Dysgenesis and Hypodontia,
Nail-Patella Syndrome, Nance-Horan Syndrome, Nanocephalic Dwarfism, Nanocephaly,
Nanophthalmia, Narcolepsy, Narcoleptic syndrome, NARP, Nasal-fronto-faciodyplasia,
Nasal Alar Hypoplasia Hypothyroidism Pancreatic Achylia Congenital Deafness,
Nasomaxillary Hypoplasia, Nasu Lipodystrophy, NBIA1, ND, NDI, NDP, Necrotizing
25 Encephalomyelopathy of Leigh's, Necrotizing Respiratory Granulomatosis, Neill-
Dingwall Syndrome, Nelson Syndrome, Nemaline myopathy, Neonatal
Adrenoleukodystrophy, Neonatal Adrenoleukodystrophy (NALD), Neonatal
Adrenoleukodystrophy (ALD), Neonatal Autosomal Recessive Polycystic Kidney Disease,
Neonatal Dwarfism, Neonatal Hepatitis, Neonatal Hypoglycemia, Neonatal Lactose
30 Intolerance, Neonatal Lymphedema due to Exudative Enteropathy, Neonatal Progeroid
Syndrome, Neonatal Pseudo-Hydrocephalic Progeroid Syndrome of Wiedemann-

- Rautenstrauch, Neoplastic Arachnoiditis, Nephroblastom, Nephrogenic Diabetes Insipidus, Nephronophthisis Familial Juvenile, Nephropathic Cystinosis, Nephropathy-Pseudohermaphroditism-Wilms Tumor, Nephrosis-Microcephaly Syndrome, Nephrosis-Neuronal Dysmigration Syndrome, Nephrotic-Glycosuric-Dwarfism-Rickets-
- 5 Hypophosphatemic Syndrome, Netherton Disease, Netherton Syndrome, Netherton Syndrome Ichthyosis, Nettleship Falls Syndrome (X-Linked), Neu-Laxova Syndrome, Neuhauser Syndrome, Neural-tube defects, Neuralgic Amyotrophy, Neuraminidase Deficiency, Neuraocutaneous melanosis, Neurinoma of the Acoustic Nerve, Neurinoma, Neuroacanthocytosis, Neuroaxonal Dystrophy Schindler Type, Neurodegeneration with
 - 10 brain iron accumulation type 1 (NBIA1), Neurofibroma of the Acoustic Nerve, Neurogenic Arthrogryposis Multiplex Congenita, Neuromyelitis Optica, Neuromyotonia, Neuromyotonia, Focal, Neuromyotonia, Generalized, Familial, Neuromyotonia, Generalized, Sporadic, Neuronal Axonal Dystrophy Schindler Type, Neuronal Ceroid Lipofuscinosis Adult Type, Neuronal Ceroid Lipofuscinosis Juvenile Type, Neuronal
 - 15 Ceroid Lipofuscinosis Type 1, Neuronopathic Acute Gaucher Disease, Neuropathic Amyloidosis, Neuropathic Beriberi, Neuropathy Ataxia and Retinitis Pigmentosa, Neuropathy of Brachialplexus Syndrome, Neuropathy Hereditary Sensory Type I, Neuropathy Hereditary Sensory Type II, Neutral Lipid Storage Disease, Nevii, Nevroid Basal Cell Carcinoma Syndrome, Nevus, Nevus Cavemosus, Nevus Comedonicus, Nevus
 - 20 Depigmentosus, Nevus Sebaceous of Jadassohn, Nezelof's Syndrome, Nezelof's Thymic Aplasia, Nezelof Type Severe Combined Immunodeficiency, NF, NF1, NF2, NF-1, NF-2, NHS, Niemann Pick Disease, Nieman Pick disease Type A (acute neuronopathic form), Nieman Pick disease Type B, Nieman Pick Disease Type C (chronic neuronopathic form), Nieman Pick disease Type D (Nova Scotia variant), Nieman Pick disease Type E, Nieman
 - 25 Pick disease Type F (sea-blue histiocyte disease), Night Blindness, Nigrospinodontatal Degeneration, Niikawakuroki Syndrome, NLS, NM, Noack Syndrome Type I, Nocturnal Myoclonus Hereditary Essential Myoclonus, Nodular Cornea Degeneration, Non-Bullous CIE, Non-Bullous Congenital Ichthyosiform Erythroderma, Non-Communicating Hydrocephalus, Non-Deletion Type Alpha-Thalassemia / Mental Retardation syndrome,
 - 30 Non-Ketonic Hyperglycinemia Type I (NKHI), Non-Ketotic Hyperglycinemia, Non-Lipid Reticuloendotheliosis, Non-Neuronopathic Chronic Adult Gaucher Disease, Non-Scarring

- Epidermolysis Bullosa, Nonarteriosclerotic Cerebral Calcifications, Nonarticular Rheumatism, Noncerebral, Juvenile Gaucher Disease, Nondiabetic Glycosuria, Nonischemic Cardiac myopathy, Nonketotic Hypoglycemia and Carnitine Deficiency due to MCAD Deficiency, Nonketotic Hypoglycemia Caused by Deficiency of Acyl-CoA
- 5 Dehydrogenase, Nonketotic Glycinemia, Nonne's Syndrome, Nonne-Milroy-Meige Syndrome, Nonopalescent Opalescent Dentine, Nonpuerperal Galactorrhea-Amenorrhea, Nonsecretory Myeloma, Nonspherocytic Hemolytic Anemia, Nontropical Sprue, Noonan Syndrome, Norepinephrine, Normal Pressure Hydrocephalus, Norman-Roberts Syndrome, Norrbottnian Gaucher Disease, Norrie Disease, Norwegian Type Hereditary Cholestasis,
 - 10 NPD, NPS, NS, NSA, Nuchal Dystonia Dementia Syndrome, Nutritional Neuropathy, Nyhan Syndrome, OAV Spectrum, Obstructive Apnea, Obstructive Hydrocephalus, Obstructive Sleep Apnea, OCC Syndrome, Occlusive Thromboangiopathy, OCCS, Occult Intracranial Vascular Malformations, Occult Spinal Dysraphism Sequence, Ochoa Syndrome, Ochronosis, Ochronotic Arthritis, OCR, OCRL, Octocephaly, Ocular Albinism,
 - 15 Ocular Herpes, Ocular Myasthenia Gravis, Oculo-Auriculo-Vertebral Dysplasia, Oculo-Auriculo-Vertebral Spectrum, Oculo-Bucco-Genital Syndrome, Oculocerebral Syndrome with Hypopigmentation, Oculocerebrocutaneous Syndrome, Oculo-Cerebro-Renal, Oculocerebrorenal Dystrophy, Oculocerebrorenal Syndrome, Oculocraniosomatic Syndrome (obsolete), Oculocutaneous Albinism, Oculocutaneous Albinism Chediak-
 - 20 Higashi Type, Oculo-Dento-Digital Dysplasia, Oculodentodigital Syndrome, Oculo-Dento- Osseous Dysplasia, Oculo Gastrointestinal Muscular Dystrophy, Oculo Gastrointestinal Muscular Dystrophy, Oculomandibulodyscephaly with hypotrichosis, Oculomandibulofacial Syndrome, Oculomotor with Congenital Contractures and Muscle Atrophy, Oculosympathetic Palsy, ODD Syndrome, ODOD, Odontogenic Tumor,
 - 25 Odontotrichomelic Syndrome, OFD, OFD Syndrome, Ohio Type Amyloidosis (Type VII), OI, OI Congenita, OI Tarda, Oldfield Syndrome, Oligohydramnios Sequence, Oligophrenia Microphthalmos, Oligophrenic Polydystrophy, Olivopontocerebellar Atrophy, Olivopontocerebellar Atrophy with Dementia and Extrapyrimal Signs, Olivopontocerebellar Atrophy with Retinal Degeneration, Olivopontocerebellar Atrophy I,
 - 30 Olivopontocerebellar Atrophy II, Olivopontocerebellar Atrophy III, Olivopontocerebellar Atrophy IV, Olivopontocerebellar Atrophy V, Ollier Disease, Ollier Osteochondromatosis,

Omphalocele-Visceromegaly-Macroglossia Syndrome, Ondine's Curse, Onion-Bulb
 Neuropathy, Onion Bulb Polyneuropathy, Onychoosteodysplasia, Onychotrichodysplasia
 with Neutropenia, OPCA, OPCA I, OPCA II, OPCA III, OPCA IV, OPCA V, OPD
 Syndrome, OPD Syndrome Type I, OPD Syndrome Type II, OPD I Syndrome, OPD II
 5 Syndrome, Ophthalmoarthropathy, Ophthalmoplegia-Intestinal Pseudoobstruction,
 Ophthalmoplegia, Pigmentary Degeneration of the Retina and Cardio myopathy,
 Ophthalmoplegia Plus Syndrome, Ophthalmoplegia Syndrome, Opitz BBB Syndrome,
 Opitz BBB/G Compound Syndrome, Opitz BBBG Syndrome, Opitz-Frias Syndrome,
 Opitz G Syndrome, Opitz G/BBB Syndrome, Opitz Hypertelorism-Hypospadias
 10 Syndrome, Opitz-Kaveggia Syndrome, Opitz Oculogenitolarhyngal Syndrome, Opitz
 Trignocephaly Syndrome, Opitz Syndrome, Opsoclonus, Opsoclonus-Myoclonus,
 Ophthalmoneuromyelitis, Optic Atrophy Polyneuropathy and Deafness, Optic
 Neuroencephalomyelopathy, Optic Neuromyelitis, Opticomyelitis, Optochiasmatic
 Arachnoiditis, Oral-Facial Clefts, Oral-facial Dyskinesia, Oral Facial Dystonia, Oral-
 15 Facial-Digital Syndrome, Oral-Facial-Digital Syndrome Type I, Oral-Facial-Digital
 Syndrome I, Oral-Facial-Digital Syndrome II, Oral-Facial-Digital Syndrome III, Oral-
 Facial-Digital Syndrome IV, Orbital Cyst with Cerebral and Focal Dermal Malformations,
 Ornithine Carbamyl Transferase Deficiency, Ornithine Transcarbamylase Deficiency,
 Orocraniodigital Syndrome, Orofaciodigital Syndrome, Oromandibular Dystonia,
 20 Orthostatic Hypotension, Osler-Weber-Rendu disease, Osseous-Oculo-Dento Dysplasia,
 Osseous-Oculo-Dento Dysplasia, Osteitis deformans, Osteochondrodystrophy Deformans,
 Osteochondroplasia, Osteodysplasty of Melnick and Needles, Osteogenesis Imperfect,
 Osteogenesis Imperfecta, Osteogenesis Imperfecta Congenita, Osteogenesis Imperfecta
 Tarda, Osteohypertrophic Nevus Flammeus, Osteopathia Hyperostotica Scleroticans
 25 Multiplex Infantilis, Osteopathia Hyperostotica Scleroticans Multiplex Infantilis,
 Osteopathyrosis, Osteopetrosis, Osteopetrosis Autosomal Dominant Adult Type,
 Osteopetrosis Autosomal Recessive Malignant Infantile Type, Osteopetrosis Mild
 Autosomal Recessive Intermediate Typ, Osteosclerosis Fragilis Generalisata,
 Osteosclerotic Myeloma, Ostium Primum Defect (endocardial cushion defects included),
 30 Ostium Secundum Defect, OTC Deficiency, Oto Palato Digital Syndrome, Oto-Palato-
 Digital Syndrome Type I, Oto-Palatal-Digital Syndrome Type II, Otodental Dysplasia,

- Otopalatodigital Syndrome, Otopalatodigital Syndrome Type II, Oudtshoorn Skin, Ovarian Dwarfism Turner Type, Ovary Aplasia Turner Type, OWR, Oxalosis, Oxidase deficiency, Oxycephaly, Oxycephaly-Acrocephaly, P-V, PA, PAC, Pachyonychia Ichtyosiforme, Pachyonychia Congenita with Natal Teeth, Pachyonychia Congenita,
- 5 Pachyonychia Congenita Keratosis Disseminata Circumscripta (follicularis), Pachyonychia Congenita Jadassohn-Lewandowsky Type, PAF with MSA, Paget's Disease, Paget's Disease of Bone, Paget's Disease of the Breast, Paget's Disease of the Nipple, Paget's Disease of the Nipple and Areola, Pagon Syndrome, Painful Ophthalmoplegia, PAIS, Palatal Myoclonus, Palato-Oto-Digital Syndrome, Palatal-Oto-Digital Syndrome Type I,
- 10 Palatal-Oto-Digital Syndrome Type II, Pallister Syndrome, Pallister-Hall Syndrome, Pallister-Killian Mosaic Syndrome, Pallister Mosaic Aneuploidy, Pallister Mosaic Syndrome, Pallister Mosaic Syndrome Tetrasomy 12p, Pallister-W Syndrome, Palmoplantar Hyperkeratosis and Alopecia, Palsy, Pancreatic Fibrosis, Pancreatic Insufficiency and Bone Marrow Dysfunction, Pancreatic Ulcerogenic Tumor Syndrome,
- 15 Panmyelophthisis, Panmyelopathy, Pantothenate kinase associated neurodegeneration (PKAN), Papillon-Lefevre Syndrome, Papillotonic Psuedotabes, Paralysis Periodica Paramyotonia, Paralytic Beriberi, Paralytic Brachial Neuritis, Paramedian Lower Lip Pits-Popliteal Pyerygium Syndrome, Paramedian Diencephalic Syndrome, Paramyeloidosis, Paramyoclonus Multiple, Paramyotonia Congenita, Paramyotonia Congenita of Von
- 20 Eulenburg, Parkinson's disease, Paroxysmal Atrial Tachycardia, Paroxysmal Cold Hemoglobinuria, Paroxysmal Dystonia, Paroxysmal Dystonia Choreathetosis, Paroxysmal Kinesigenic Dystonia, Paroxysmal Nocturnal Hemoglobinuria, Paroxysmal Normal Hemoglobinuria, Paroxysmal Sleep, Parrot Syndrome, Parry Disease, Parry-Romberg Syndrome, Parsonage-Turner Syndrome, Partial Androgen Insensitivity Syndrome, Partial
- 25 Deletion of the Short Arm of Chromosome 4, Partial Deletion of the Short Arm of Chromosome 5, Partial Deletion of Short Arm of Chromosome 9, Partial Duplication 3q Syndrome, Partial Duplication 15q Syndrome, Partial Facial Palsy With Urinary Abnormalities, Partial Gigantism of Hands and Feet- Nevi-Hemihypertrophy-Macrocephaly, Partial Lipodystrophy, Partial Monosomy of Long Arm of Chromosome
- 30 11, Partial Monosomy of the Long Arm of Chromosome 13, Partial Spinal Sensory Syndrome, Partial Trisomy 11q, Partington Syndrome, PAT, Patent Ductus Arteriosus,

Pathological Myoclonus, Pauciarticular-Onset Juvenile Arthritis, Paulitis, PBC, PBS, PC
 Deficiency, PC Deficiency Group A, PC Deficiency Group B, PC, Eulenburg Disease,
 PCC Deficiency, PCH, PCLD, PCT, PD, PDA, PDH Deficiency, Pearson Syndrome
 5 Pyruvate Carboxylase Deficiency, Pediatric Obstructive Sleep Apnea, Peeling Skin
 Syndrome, Pelizaeus-Merzbacher Disease, Pelizaeus-Merzbacher Brain Sclerosis,
 Pellagra-Cerebellar Ataxia-Renal Aminoaciduria Syndrome, Pelvic Pain Syndrome,
 Pemphigus Vulgaris, Pena Shokeir II Syndrome, Pena Shokeir Syndrome Type II, Penile
 Fibromatosis, Penile Fibrosis, Penile Induration, Penta X Syndrome, Pentalogy of Cantrell,
 Pentalogy Syndrome, Pentasomy X, PEPCK Deficiency, Pepper Syndrome, Perheentupa
 10 Syndrome, Periarticular Fibrositis, Pericardial Constriction with Growth Failure,
 Pericollagen Amyloidosis, Perinatal Polycystic Kidney Diseases, Perineal Anus, Periodic
 Amyloid Syndrome, Periodic Peritonitis Syndrome, Periodic Somnolence and Morbid
 Hunger, Periodic Syndrome, Peripheral Cystoid Degeneration of the Retina, Peripheral
 Dysostosis-Nasal Hypoplasia-Mental Retardation, Peripheral Neuritis, Peripheral
 15 Neuropathy, Peritoneopericardial Diaphragmatic Hernia, Pernicious Anemia, Peromelia
 with Micrognathia, Peroneal Muscular Atrophy, Peroneal Nerve Palsy, Peroutka Sneeze,
 Peroxisomal Acyl-CoA Oxidase, Peroxisomal Beta-Oxidation Disorders, Peroxisomal
 Bifunctional Enzyme, Peroxisomal Thiolase, Peroxisomal Thiolase Deficiency, Persistent
 Truncus Arteriosus, Perthes Disease, Petit Mal Epilepsy, Petit Mal Variant, Peutz-Jeghers
 20 Syndrome, Peutz-Touraine Syndrome, Peyronie Disease, Pfeiffer, Pfeiffer Syndrome Type
 I, PGA I, PGA II, PGA III, PGK, PH Type I, PH Type I, Pharyngeal Pouch Syndrome,
 PHD Short-Chain Acyl-CoA Dehydrogenase Deficiency, Phenylalanine Hydroxylase
 Deficiency, Phenylalaninemia, Phenylketonuria, Phenylpyruvic Oligophrenia, Phocomelia,
 Phocomelia Syndrome, Phosphoenolpyruvate Carboxykinase Deficiency,
 25 Phosphofructokinase Deficiency, Phosphoglycerate Kinase Deficiency,
 Phosphoglycerokinase, Phosphorylase 6 Kinase Deficiency, Phosphorylase Deficiency
 Glycogen Storage Disease, Phosphorylase Kinase Deficiency of Liver, Photic Sneeze
 Reflex, Photic Sneezing, Phototherapeutic keratectomy, PHS, Physicist John Dalton,
 Phytanic Acid Storage Disease, Pi Phenotype ZZ, PI, Pick Disease of the Brain, Pick's
 30 Disease, Pickwickian Syndrome, Pierre Robin Anomalad, Pierre Robin Complex, Pierre
 Robin Sequence, Pierre Robin Syndrome, Pierre Robin Syndrome with Hyperphalangy and

- Clinodactyly, Pierre-Marie's Disease, Pigmentary Degeneration of Globus Pallidus
 Substantia Nigra Red Nucleus, Pili Torti and Nerve Deafness, Pili Torti-Sensorineural
 Hearing Loss, Pituitary Dwarfism II, Pituitary Tumor after Adrenalectomy, Pityriasis
 Pilaris, Pityriasis Rubra Pilaris, PJS, PKAN, PKD, PKD1, PKD2, PKD3, PKU, PKU1,
 5 Plagiocephaly, Plasma Cell Myeloma, Plasma Cell Leukemia, Plasma Thromboplastin
 Component Deficiency, Plasma Transglutaminase Deficiency, Plastic Induration Corpora
 Cavernosa, Plastic Induration of the Penis, PLD, Plicated Tongue, PLS, PMD,
 Pneumorenal Syndrome, PNH, PNM, PNP Deficiency, POD, POH, Poikiloderma
 Atrophicans and Cataract, Poikiloderma Congenitale, Poland Anomaly, Poland Sequence,
 10 Poland Syndactyly, Poland Syndrome, Poliodystrophia Cerebri Progressiva, Polyarthritis
 Enterica, Polyarteritis Nodosa, Polyarticular-Onset Juvenile Arthritis Type I, Polyarticular-
 Onset Juvenile Arthritis Type II, Polyarticular-Onset Juvenile Arthritis Types I and II,
 Polychondritis, Polycystic Kidney Disease, Polycystic Kidney Disease Medullary Type,
 Polycystic Liver Disease, Polycystic Ovary Disease, Polycystic Renal Diseases,
 15 Polydactyly-Joubert Syndrome, Polydysplastic Epidermolysis Bullosa, Polydystrophia
 Oligophrenia, Polydystrophic Dwarfism, Polyglandular Autoimmune Syndrome Type III,
 Polyglandular Autoimmune Syndrome Type II, Polyglandular Autoimmune Syndrome
 Type I, Polyglandular Autoimmune Syndrome Type II, Polyglandular Deficiency
 Syndrome Type II, Polyglandular Syndromes, Polymorphic Macula Lutea Degeneration,
 20 Polymorphic Macular Degeneration, Polymorphism of Platelet Glycoprotein Ib,
 Polymorphous Corneal Dystrophy Hereditary, Polymyalgia Rheumatica, Polymyositis and
 Dermatomyositis, Primary Agammaglobulinemia, Polyneuritis Peripheral,
 Polyneuropathy-Deafness-Optic Atrophy, Polyneuropathy Peripheral, Polyneuropathy and
 Polyradiculoneuropathy, Polyostotic Fibrous Dysplasia, Polyostotic Sclerosing
 25 Histiocytosis, Polyposis Familial, Polyposis Gardner Type, Polyposis Hamartomatous
 Intestinal, Polyposis-Osteomatosis-Epidermoid Cyst Syndrome, Polyposis Skin
 Pigmentation Alopecia and Fingernail Changes, Polyps and Spots Syndrome, Polyserositis
 Recurrent, Polysomy Y, Polysyndactyly with Peculiar Skull Shape, Polysyndactyly-
 Dysmorphic Craniofacies Greig Type, Pompe Disease, Pompe Disease, Popliteal
 30 Pterygium Syndrome, Porcupine Man, Porencephaly, Porencephaly, Porphobilinogen
 deaminase (PBG-D), Porphyria, Porphyria Acute Intermittent, Porphyria ALA-D,

- Porphyria Cutanea Tarda, Porphyria Cutanea Tarda Hereditaria, Porphyria Cutanea Tarda Symptomatica, Porphyria Hepatica Variegata, Porphyria Swedish Type, Porphyria Variegata, Porphyria Acute Intermittent, Porphyrins, Porphyria Decalvans, Port Wine Stains, Portuguese Type Amyloidosis, Post-Infective Polyneuritis, Postanoxic Intention
- 5 Myoclonus, Postaxial Acrofacial Dysostosis, Postaxial Polydactyly, Postencephalic Intention Myoclonus, Posterior Corneal Dystrophy Hereditary, Posterior Thalamic Syndrome, Postmyelographic Arachnoiditis, Postnatal Cerebral Palsy, Postoperative Cholestasis, Postpartum Galactorrhea-Amenorrhea Syndrome, Postpartum Hypopituitarism, Postpartum Panhypopituitary Syndrome, Postpartum Panhypopituitarism,
- 10 Postpartum Pituitary Necrosis, Postural Hypotension, Potassium-Losing Nephritis, Potassium Loss Syndrome, Potter Type I Infantile Polycystic Kidney Diseases, Potter Type III Polycystic Kidney Disease, PPH, PPS, Prader-Willi Syndrome, Prader-Labhart-Willi Fancone Syndrome, Prealbumin Tyr-77 Amyloidosis, Preexcitation Syndrome, Pregnenolone Deficiency, Premature Atrial Contractions, Premature Senility Syndrome,
- 15 Premature Supraventricular Contractions, Premature Ventricular Complexes, Prenatal or Connatal Neuroaxonal Dystrophy, Presenile Dementia, Presenile Macula Lutea Retinae Degeneration, Primary Adrenal Insufficiency, Primary Agammaglobulinemias, Primary Aldosteronism, Primary Alveolar Hypoventilation, Primary Amyloidosis, Primary Anemia, Primary Beriberi, Primary Biliary, Primary Biliary Cirrhosis, Primary Brown Syndrome,
- 20 Primary Carnitine Deficiency, Primary Central Hypoventilation Syndrome, Primary Ciliary Dyskinesia Kartagener Type, Primary Cutaneous Amyloidosis, Primary Dystonia, Primary Failure Adrenocortical Insufficiency, Primary Familial Hypoplasia of the Maxilla, Primary Hemochromatosis, Primary Hyperhidrosis, Primary Hyperoxaluria [Type I], Primary Hyperoxaluria Type 1 (PH1), Primary Hyperoxaluria Type 1, Primary
- 25 Hyperoxaluria Type II, Primary Hyperoxaluria Type III, Primary Hypogonadism, Primary Intestinal Lymphangiectasia, Primary Lateral Sclerosis, Primary Nonhereditary Amyloidosis, Primary Obliterative Pulmonary Vascular Disease, Primary Progressive Multiple Sclerosis, Primary Pulmonary Hypertension, Primary Reading Disability, Primary Renal Glycosuria, Primary Sclerosing Cholangitis, Primary Thrombocythemia, Primary
- 30 Tumors of Central Nervous System, Primary Visual Agnosia, Proctocolitis Idiopathic, Proctocolitis Idiopathic, Progeria of Adulthood, Progeria of Childhood, Progeroid Nanism,

- Progeroid Short Stature with Pigmented Nevi, Progeroid Syndrome of De Barsy, Progressive Autonomic Failure with Multiple System Atrophy, Progressive Bulbar Palsy, Progressive Bulbar Palsy Included, Progressive Cardiomyopathic Lentiginosis, Progressive Cerebellar Ataxia Familial, Progressive Cerebral Poliodystrophy, Progressive Choroidal
- 5 Atrophy, Progressive Diaphyseal Dysplasia, Progressive Facial Hemiatrophy, Progressive Familial Myoclonic Epilepsy, Progressive Hemifacial Atrophy, Progressive Hypoerythemia, Progressive Infantile Poliodystrophy, Progressive Lenticular Degeneration, Progressive Lipodystrophy, Progressive Muscular Dystrophy of Childhood, Progressive Myoclonic Epilepsy, Progressive Osseous Heteroplasia, Progressive Pallid
- 10 Degeneration Syndrome, Progressive Spinobulbar Muscular Atrophy, Progressive Supranuclear Palsy, Progressive Systemic Sclerosis, Progressive Tapetochoroidal Dystrophy, Proline Oxidase Deficiency, Propionic Acidemia, Propionic Acidemia Type I (PCCA Deficiency), Propionic Acidemia Type II (PCCB Deficiency), Propionyl CoA Carboxylase Deficiency, Protanomaly, Protanopia, Protein-Losing Enteropathy Secondary
- 15 to Congestive Heart Failure, Proteus Syndrome, Proximal Deletion of 4q Included, PRP, PRS, Prune Belly Syndrome, PS, Pseudo-Hurler Polydystrophy, Pseudo-Polydystrophy, Pseudoacanthosis Nigricans, Pseudoachondroplasia, Pseudocholinesterase Deficiency, Pseudogout Familial, Pseudothrombophilia, Pseudohermaphroditism, Pseudohermaphroditism-Nephron Disorder-Wilm's Tumor, Pseudohypertrophic Muscular
- 20 Dystrophy, Pseudohypoparathyroidism, Pseudohypophosphatasia, Pseudopolydystrophy, Pseudothalidomide Syndrome, Pseudoxanthoma Elasticum, Psoriasis, Psorospermosis Follicularis, PSP, PSS, Psychomotor Convulsion, Psychomotor Epilepsy, Psychomotor Equivalent Epilepsy, PTC Deficiency, Pterygium, Pterygium Colli Syndrome, Pterygium Universale, Pterygolympangiectasia, Pulmonary Atresia, Pulmonary
- 25 Lymphangiomyomatosis, Pulmonary Stenosis, Pulmonic Stenosis-Ventricular Septal Defect, Pulp Stones, Pulpal Dysplasia, Pulseless Disease, Pure A lymphocytosis, Pure Cutaneous Histiocytosis, Purine Nucleoside Phosphorylase Deficiency, Purpura Hemorrhagica, Purtilo Syndrome, PXE, PXE Dominant Type, PXE Recessive Type, Pycnodysostosis, Pyknodysostosis, Pyknoepilepsy, Pyroglutamic Aciduria,
- 30 Pyroglutamicaciduria, Pyrroline Carboxylate Dehydrogenase Deficiency, Pyruvate Carboxylase Deficiency, Pyruvate Carboxylase Deficiency Group A, Pyruvate

- Carboxylase Deficiency Group B, Pyruvate Dehydrogenase Deficiency, Pyruvate Kinase Deficiency, q25-qter, q26 or q27-qter, q31 or 32-qter, QT Prolongation with Extracellular Hypohypocalcemia, QT Prolongation without Congenital Deafness, QT Prolonged with Congenital Deafness, Quadripareisis of Cerebral Palsy, Quadriplegia of Cerebral Palsy,
- 5 Quantal Squander, Quantal Squander, r4, r6, r14, r 18, r21, r22, Rachischisis Posterior, Radial Aplasia-Amegakaryocytic Thrombocytopenia, Radial Aplasia-Thrombocytopenia Syndrome, Radial Nerve Palsy, Radicular Neuropathy Sensory, Radicular Neuropathy Sensory Recessive, Radicular Dentin Dysplasia, Rapid-onset Dystonia-parkinsonism, Rapp-Hodgkin Syndrome, Rapp-Hodgkin (hypohidrotic) Ectodermal Dysplasia syndrome,
- 10 Rapp-Hodgkin Hypohidrotic Ectodermal Dysplasias, Rare hereditary ataxia with polyneuritic changes and deafness caused by a defect in the enzyme phytanic acid hydroxylase, Rautenstrauch-Wiedemann Syndrome, Rautenstrauch-Wiedemann Type Neonatal Progeria, Raynaud's Phenomenon, RDP, Reactive Functional Hypoglycemia, Reactive Hypoglycemia Secondary to Mild Diabetes, Recessive Type Kenny-Caffe
- 15 Syndrome, Recklin Recessive Type Myotonia Congenita, Recklinghausen Disease, Rectoperineal Fistula, Recurrent Vomiting, Reflex Neurovascular Dystrophy, Reflex Sympathetic Dystrophy Syndrome, Refractive Errors, Refractory Anemia, Refrigeration Palsy, Refsum Disease, Refsum's Disease, Regional Enteritis, Reid-Barlow's syndrome, Reifenstein Syndrome, Reiger Anomaly-Growth Retardation, Reiger Syndrome, Reimann
- 20 Periodic Disease, Reimann's Syndrome, Reis-Bucklers Corneal Dystrophy, Reiter's Syndrome, Relapsing Guillain-Barre Syndrome, Relapsing-Remitting Multiple Sclerosis, Renal Agenesis, Renal Dysplasia-Blindness Hereditary, Renal Dysplasia-Retinal Aplasia Loken-Senior Type, Renal Glycosuria, Renal Glycosuria Type A, Renal Glycosuria Type B, Renal Glycosuria Type O, Renal-Oculocerebrodystrophy, Renal-Retinal Dysplasia with
- 25 Medullary Cystic Disease, Renal-Retinal Dystrophy Familial, Renal-Retinal Syndrome, Rendu-Osler-Weber Syndrome, Respiratory Acidosis, Respiratory Chain Disorders, Respiratory Myoclonus, Restless Legs Syndrome, Restrictive Cardio myopathy, Retention Hyperlipemia, Rethore Syndrome (obsolete), Reticular Dysgenesis, Retinal Aplastic-Cystic Kidneys-Joubert Syndrome, Retinal Cone Degeneration, Retinal Cone Dystrophy,
- 30 Retinal Cone-Rod Dystrophy, Retinitis Pigmentosa, Retinitis Pigmentosa and Congenital Deafness, Retinoblastoma, Retinol Deficiency, Retinoschisis, Retinoschisis Juvenile,

Retraction Syndrome, Retrobulbar Neuropathy, Retrolenticular Syndrome, Rett Syndrome, Reverse Coarction, Reye Syndrome, Reye's Syndrome, RGS, Rh Blood Factors, Rh Disease, Rh Factor Incompatibility, Rh Incompatibility, Rhesus Incompatibility, Rheumatic Fever, Rheumatoid Arthritis, Rheumatoid Myositis, Rhinosinusogenic Cerebral

5 Arachnoiditis, Rhizomelic Chondrodysplasia Punctata (RCDP), Acatalasemia, Classical Refsum disease, RHS, Rhythmical Myoclonus, Rib Gap Defects with Micrognathia, Ribbing Disease (obsolete), Ribbing Disease, Richner-Hanhart Syndrome, Rieger Syndrome, Rieter's Syndrome, Right Ventricular Fibrosis, Riley-Day Syndrome, Riley-Smith syndrome, Ring Chromosome 14, Ring Chromosome 18, Ring 4, Ring 4

10 Chromosome, Ring 6, Ring 6 Chromosome, Ring 9, Ring 9 Chromosome R9, Ring 14, Ring 15, Ring 15 Chromosome (mosaic pattern), Ring 18, Ring Chromosome 18, Ring 21, Ring 21 Chromosome, Ring 22, Ring 22 Chromosome, Ritter Disease, Ritter-Lyell Syndrome, RLS, RMSS, Roberts SC-Phocomelia Syndrome, Roberts Syndrome, Roberts Tetraphocomelia Syndrome, Robertson's Ectodermal Dysplasias, Robin Anomalad, Robin

15 Sequence, Robin Syndrome, Robinow Dwarfism, Robinow Syndrome, Robinow Syndrome Dominant Form, Robinow Syndrome Recessive Form, Rod myopathy, Roger Disease, Rokitansky's Disease, Romano-Ward Syndrome, Romberg Syndrome, Rotters Teeth, Rosenberg-Chutorian Syndrome, Rosewater Syndrome, Rosselli-Gulienatti Syndrome, Rothmund-Thomson Syndrome, Roussy-Levy Syndrome, RP, RS X-Linked,

20 RS, RSDS, RSH Syndrome, RSS, RSTS, RTS, Rubella Congenital, Rubinstein Syndrome, Rubinstein-Taybi Syndrome, Rubinstein Taybi Broad Thumb-Hallux syndrome, Rufous Albinism, Ruhr's Syndrome, Russell's Diencephalic Cachexia, Russell's Syndrome, Russell Syndrome, Russell-Silver Dwarfism, Russell-Silver Syndrome, Russell-Silver Syndrome X-linked, Ruvalcaba-Myhre-Smith syndrome (RMSS), Ruvalcaba Syndrome,

25 Ruvalcaba Type Osseous Dysplasia with Mental Retardation, Sacral Regression, Sacral Agenesis Congenital, SAE, Saethre-Chotzen Syndrome, Sakati, Sakati Syndrome, Sakati-Nyhan Syndrome, Salaam Spasms, Salivosudoriparous Syndrome, Salzman Nodular Corneal Dystrophy, Sandhoff Disease, Sanfilippo Syndrome, Sanfilippo Type A, Sanfilippo Type B, Santavuori Disease, Santavuori-Haltia Disease, Sarcoid of Boeck,

30 Sarcoidosis, Sathre-chotzen, Saturday Night Palsy, SBMA, SC Phocomelia Syndrome, SC Syndrome, SCA 3, SCAD Deficiency, SCAD Deficiency Adult-Onset Localized, SCAD

- Deficiency Congenital Generalized, SCAD, SCADH Deficiency, Scalded Skin Syndrome, Scalp Defect Congenital, Scaphocephaly, Scapula Elevata, Scapuloperoneal myopathy, Scapuloperoneal Muscular Dystrophy, Scapuloperoneal Syndrome Myopathic Type, Scarring Bullosa, SCHAD, Schaumann's Disease, Scheie Syndrome, Schereshevskii-Turner
- 5 Syndrome, Schilder Disease, Schilder Encephalitis, Schilder's Disease, Schindler Disease Type I (Infantile Onset), Schindler Disease Infantile Onset, Schindler Disease, Schindler Disease Type II (Adult Onset), Schinzel Syndrome, Schinzel-Giedion Syndrome, Schinzel Acrocallosal Syndrome, Schinzel-Giedion Midface-Retraktion Syndrome, Schizencephaly, Schmid Type Metaphyseal Chondrodysplasia, Schmid Metaphyseal Dysostosis, Schmid-
- 10 Fraccaro Syndrome, Schmidt Syndrome, Schopf-Schultz-Passarge Syndrome, Schueller-Christian Disease, Schut-Haymaker Type, Schwartz-Jampel-Aberfeld Syndrome, Schwartz-Jampel Syndrome Types 1A and 1B, Schwartz-Jampel Syndrome, Schwartz-Jampel Syndrome Type 2, SCID, Scleroderma, Sclerosis Familial Progressive Systemic, Sclerosis Diffuse Familial Brain, Scott Craniodigital Syndrome With Mental Retardation,
- 15 Scrotal Tongue, SCS, SD, SDS, SDYS, Seasonal Conjunctivitis, Sebaceous Nevus Syndrome, Sebaceous nevus, Seborrheic Keratosis, Seborrheic Warts, Seckel Syndrome, Seckel Type Dwarfism, Second Degree Congenital Heart Block, Secondary Amyloidosis, Secondary Blepharospasm, Secondary Non-tropical Sprue, Secondary Brown Syndrome, Secondary Beriberi, Secondary Generalized Amyloidosis, Secondary Dystonia, Secretory
- 20 Component Deficiency, Secretory IgA Deficiency, SED Tarda, SED Congenital, SEDC, Segmental linear achromic nevus, Segmental Dystonia, Segmental Myoclonus, Seip Syndrome, Seitelberger Disease, Seizures, Selective Deficiency of IgG Subclasses, Selective Mutism, Selective Deficiency of IgG Subclass, Selective IgM Deficiency, Selective Mutism, Selective IgA Deficiency, Self-Healing Histiocytosis, Semilobar
- 25 Holoprosencephaly, Seminiferous Tubule Dysgenesis, Senile Retinoschisis, Senile Warts, Senior-Loken Syndrome, Sensory Neuropathy Hereditary Type I, Sensory Neuropathy Hereditary Type II, Sensory Neuropathy Hereditary Type I, Sensory Radicular Neuropathy, Sensory Radicular Neuropathy Recessive Sepsis, Septic Progressive Granulomatosis, Septo-Optic Dysplasia, Serous Circumscribed Meningitis, Serum Protease
- 30 Inhibitor Deficiency, Serum Carnosinase Deficiency, Setleis Syndrome, Severe Combined Immunodeficiency, Severe Combined Immunodeficiency with Adenosine Deaminase

- Deficiency, Severe Combined Immunodeficiency (SCID), Sex Reversal, Sexual
 Infantilism, SGB Syndrome, Sheehan Syndrome, Shields Type Dentinogenesis Imperfecta,
 Shingles, varicella-zoster virus, Ship Beriberi, SHORT Syndrome, Short Arm 18 Deletion
 Syndrome, Short Chain Acyl CoA Dehydrogenase Deficiency, Short Chain Acyl-CoA
 5 Dehydrogenase (SCAD) Deficiency, Short Stature and Facial Telangiectasis, Short Stature
 Facial/Skeletal Anomalies-Retardation-Macrodontia, Short Stature-Hyperextensibility-
 Rieger Anomaly-Teething Delay, Short Stature-Onychodysplasia, Short Stature
 Telangiectatic Erythema of the Face, SHORT Syndrome, Shoshin Beriberi, Shoulder girdle
 syndrome, Shprintzen-Goldberg Syndrome, Shulman Syndrome, Shwachman-Bodian
 10 Syndrome, Shwachman-Diamond Syndrome, Shwachman Syndrome, Shwachman-
 Diamond-Oski Syndrome, Shwachmann Syndrome, Shy Drager Syndrome, Shy-Magee
 Syndrome, SI Deficiency, Sialidase Deficiency, Sialidosis Type I Juvenile, Sialidosis Type
 II Infantile, Sialidosis, Sialolipidosis, Sick Sinus Syndrome, Sickle Cell Anemia, Sickle
 Cell Disease, Sickle Cell-Hemoglobin C Disease, Sickle Cell-Hemoglobin D Disease,
 15 Sickle Cell-Thalassemia Disease, Sickle Cell Trait, Sideroblastic Anemias, Sideroblastic
 Anemia, Sideroblastosis, SIDS, Siegel-Cattan-Mamou Syndrome, Siemens-Bloch type
 Pigmented Dermatoses, Siemens Syndrome, Siewerling-Creutzfeldt Disease, Siewert
 Syndrome, Silver Syndrome, Silver-Russell Dwarfism, Silver-Russell Syndrome,
 Simmond's Disease, Simons Syndrome, Simplex Epidermolysis Bullosa, Simpson
 20 Dysmorphia Syndrome, Simpson-Golabi-Behmel Syndrome, Sinding-Larsen-Johansson
 Disease, Singleton-Merten Syndrome, Sinus Arrhythmia, Sinus Venosus, Sinus
 tachycardia, Sirenomelia Sequence, Sirenomelus, Situs Inversus Bronchiectasis and
 Sinusitis, SJA Syndrome, Sjogren Larsson Syndrome Ichthyosis, Sjogren Syndrome,
 Sjögren's Syndrome, SJS, Skeletal dysplasia, Skeletal Dysplasia Weismann Netter Stuhl
 25 Type, Skin Peeling Syndrome, Skin Neoplasms, Skull Asymmetry and Mild Retardation,
 Skull Asymmetry and Mild Syndactyly, SLE, Sleep Epilepsy, Sleep Apnea, SLO, Sly
 Syndrome, SMA, SMA Infantile Acute Form, SMA I, SMA III, SMA type I, SMA type II,
 SMA type III, SMA3, SMAX1, SMCR, Smith Lemli Opitz Syndrome, Smith Magenis
 Syndrome, Smith-Magenis Chromosome Region, Smith-McCort Dwarfism, Smith-Opitz-
 30 Inborn Syndrome, Smith Disease, Smoldering Myeloma, SMS, SNE, Sneezing From Light
 Exposure, Sodium valproate, Solitary Plasmacytoma of Bone, Sorsby Disease, Sotos

- Syndrome, Souques-Charcot Syndrome, South African Genetic Porphyria, Spasmodic Dysphonia, Spasmodic Torticollis, Spasmodic Wryneck, Spastic Cerebral Palsy, Spastic Colon, Spastic Dysphonia, Spastic Paraplegia, SPD Calcinosis, Specific Antibody Deficiency with Normal Immunoglobulins, Specific Reading Disability, SPH2,
- 5 Spherocytic Anemia, Spherocytosis, Spherophakia-Brachymorphia Syndrome, Sphingomyelin Lipidosis, Sphingomyelinase Deficiency, Spider fingers, Spielmeyer-Vogt Disease, Spielmeyer-Vogt-Batten Syndrome, Spina Bifida, Spina Bifida Aperta, Spinal Arachnoiditis, Spinal Arteriovenous Malformation, Spinal Ataxia Hereditofamilial, Spinal and Bulbar Muscular Atrophy, Spinal Diffuse Idiopathic Skeletal Hyperostosis, Spinal
- 10 DISH, Spinal Muscular Atrophy, Spinal Muscular Atrophy All Types, Spinal Muscular Atrophy Type ALS, Spinal Muscular Atrophy-Hypertrophy of the Calves, Spinal Muscular Atrophy Type I, Spinal Muscular Atrophy Type III, Spinal Muscular Atrophy type 3, Spinal Muscular Atrophy-Hypertrophy of the Calves, Spinal Ossifying Arachnoiditis, Spinal Stenosis, Spino Cerebellar Ataxia, Spinocerebellar Atrophy Type I, Spinocerebellar
- 15 Ataxia Type I (SCA1), Spinocerebellar Ataxia Type II (SCAII), Spinocerebellar Ataxia Type III (SCAIII), Spinocerebellar Ataxia Type III (SCA 3), Spinocerebellar Ataxia Type IV (SCAIV), Spinocerebellar Ataxia Type V (SCAV), Spinocerebellar Ataxia Type VI (SCAVI), Spinocerebellar Ataxia Type VII (SCAVII), Spirochetal Jaundice, Splenic Agenesis Syndrome, Splenic Ptosis, Splenoptosis, Split Hand Deformity-Mandibulofacial
- 20 Dysostosis, Split Hand Deformity, Spondyloarthritis, Spondylocostal Dysplasia - Type I, Spondyloepiphyseal Dysplasia Tarda, Spondylothoracic Dysplasia, Spondylotic Caudal Radiculopathy, Sponge Kidney, Spongioblastoma Multiforme, Spontaneous Hypoglycemia, Sprengel Deformity, Spring Ophthalmia, SRS, ST, Stale Fish Syndrome, Staphylococcal Scalded Skin Syndrome, Stargardt's Disease, Startle Disease, Status
- 25 Epilepticus, Steele-Richardson-Olszewski Syndrome, Steely Hair Disease, Stein-Leventhal Syndrome, Steinert Disease, Stengel's Syndrome, Stengel-Batten-Mayou-Spielmeyer-Vogt-Stock Disease, Stenosing Cholangitis, Stenosis of the Lumbar Vertebral Canal, Stenosis, Steroid Sulfatase Deficiency, Stevanovic's Ectodermal Dysplasias, Stevens Johnson Syndrome, STGD, Stickler Syndrome, Stiff-Man Syndrome, Stiff Person
- 30 Syndrome, Still's Disease, Stilling-Turk-Duane Syndrome, Still's Disease, Stimulus-Sensitive Myoclonus, Stone Man Syndrome, Stone Man, Streeter Anomaly, Striatonigral

- Degeneration Autosomal Dominant Type, Striopallidodentate Calcinosis, Stroma, Descemet's Membrane, Stromal Corneal Dystrophy, Struma Lymphomatosa, Sturge-Kalischer-Weber Syndrome, Sturge Weber Syndrome, Sturge-Weber Phakomatosis, Subacute Necrotizing Encephalomyelopathy, Subacute Spongiform Encephalopathy,
- 5 Subacute Necrotizing Encephalopathy, Subacute Sarcoidosis, Subacute Neuronopathic, Subaortic Stenosis, Subcortical Arteriosclerotic Encephalopathy, Subendocardial Sclerosis, Succinylcholine Sensitivity, Sucrase-Isomaltase Deficiency Congenital, Sucrose-Isomaltose Malabsorption Congenital, Sucrose Intolerance Congenital, Sudanophilic Leukodystrophy ADL, Sudanophilic Leukodystrophy Pelizaeus-Merzbacher Type,
- 10 Sudanophilic Leukodystrophy Included, Sudden Infant Death Syndrome, Sudeck's Atrophy, Sugio-Kajii Syndrome, Summerskill Syndrome, Summit Acrocephalosyndactyly, Summitt's Acrocephalosyndactyly, Summitt Syndrome, Superior Oblique Tendon Sheath Syndrome, Suprarenal glands, Supravalvular Aortic Stenosis, Supraventricular tachycardia, Surdicardiac Syndrome, Surdocardiac Syndrome, SVT, Sweat Gland Abscess, Sweating
- 15 Gustatory Syndrome, Sweet Syndrome, Swiss Cheese Cartilage Syndrome, Syndactylic Oxycephaly, Syndactyly Type I with Microcephaly and Mental Retardation, Syndromatic Hepatic Ductular Hypoplasia, Syringomyelia, Systemic Aleukemic Reticuloendotheliosis, Systemic Amyloidosis, Systemic Carnitine Deficiency, Systemic Elastorrhexis, Systemic Lupus Erythematosus, Systemic Mast Cell Disease, Systemic Mastocytosis, Systemic-
- 20 Onset Juvenile Arthritis, Systemic Sclerosis, Systopic Spleen, T-Lymphocyte Deficiency, Tachyalimentation Hypoglycemia, Tachycardia, Takahara syndrome, Takayasu Disease, Takayasu Arteritis, Talipes Calcaneus, Talipes Equinovarus, Talipes Equinus, Talipes Varus, Talipes Valgus, Tandem Spinal Stenosis, Tangier Disease, Tapetoretinal Degeneration, TAR Syndrome, Tardive Dystonia, Tardive Muscular Dystrophy, Tardive
- 25 Dyskinesia, Tardive Oral Dyskinesia, Tardive Dystonia, Tardy Ulnar Palsy, Target Cell Anemia, Tarsomegaly, Tarui Disease, TAS Midline Defects Included, TAS Midline Defect, Tay Sachs Sphingolipidosis, Tay Sachs Disease, Tay Syndrome Ichthyosis, Tay Sachs Sphingolipidosis, Tay Syndrome Ichthyosis, Taybi Syndrome Type I, Taybi Syndrome, TCD, TCOF1, TCS, TD, TDO Syndrome, TDO-I, TDO-II, TDO-III,
- 30 Telangiectasis, Telecanthus with Associated Abnormalities, Telecanthus-Hypospadias Syndrome, Temporal Lobe Epilepsy, Temporal Arteritis/Giant Cell Arteritis, Temporal

- Arteritis, TEN, Tendon Sheath Adherence Superior Obliqu, Tension Myalgia, Terminal Deletion of 4q Included, Terrian Corneal Dystrophy, Teschler-Nicola/Killian Syndrome, Tethered Spinal Cord Syndrome, Tethered Cord Malformation Sequence, Tethered Cord Syndrome, Tethered Cervical Spinal Cord Syndrome, Tetrahydrobiopterin Deficiencies,
- 5 Tetrahydrobiopterin Deficiencies, Tetralogy of Fallot, Tetraphocomelia-Thrombocytopenia Syndrome, Tetrasomy Short Arm of Chromosome 9, Tetrasomy 9p, Tetrasomy Short Arm of Chromosome 18, Thalamic Syndrome, Thalamic Pain Syndrome, Thalamic Hyperesthetic Anesthesia, Thalassemia Intermedia, Thalassemia Minor, Thalassemia Major, Thiamine Deficiency, Thiamine-Responsive Maple Syrup Urine Disease, Thin-
- 10 Basement-Membrane Nephropathy, Thiolas deficiency, RCDP, Acyl-CoA dihydroxyacetonephosphate acyltransferase, Third and Fourth Pharyngeal Pouch Syndrome, Third Degree Congenital (Complete) Heart Block, Thomsen Disease, Thoracic-Pelvic-Phalangeal Dystrophy, Thoracic Spinal Canal, Thoracoabdominal Syndrome, Thoracoabdominal Ectopia Cordis Syndrome, Three M Syndrome, Three-M Slender-
- 15 Boned Nanism, Thrombasthenia of Glanzmann and Naegeli, Thrombocythemia Essential, Thrombocytopenia-Absent Radius Syndrome, Thrombocytopenia-Hemangioma Syndrome, Thrombocytopenia-Absent Radii Syndrome, Thrombophilia Hereditary Due to AT III, Thrombotic Thrombocytopenic Purpura, Thromboulcerative Colitis, Thymic Dysplasia with Normal Immunoglobulins, Thymic Agenesis, Thymic Aplasia DiGeorge
- 20 Type, Thymic Hypoplasia Agammaglobulinemias Primary Included, Thymic Hypoplasia DiGeorge Type, Thymus Congenital Aplasia, Tic Douloureux, Tics, Tinel's syndrome, Tolosa Hunt Syndrome, Tonic Spasmodic Torticollis, Tonic Pupil Syndrome, Tooth and Nail Syndrome, Torch Infection, TORCH Syndrome, Torsion Dystonia, Torticollis, Total Lipodystrophy, Total anomalous pulmonary venous connection, Touraine's Aphthosis,
- 25 Tourette Syndrome, Tourette's disorder, Townes-Brocks Syndrome, Townes Syndrome, Toxic Paralytic Anemia, Toxic Epidermal Necrolysis, Toxopachyosteose Diaphysaire Tibio-Peroniere, Toxopachyosteose, Toxoplasmosis Other Agents Rubella Cytomegalovirus Herpes Simplex, Tracheoesophageal Fistula with or without Esophageal Atresia, Tracheoesophageal Fistula, Transient neonatal myasthenia gravis, Transitional
- 30 Atrioventricular Septal Defect, Transposition of the great arteries, Transtelephonic Monitoring, Transthyretin Methionine-30 Amyloidosis (Type I), Trapezoidocephaly-

- Multiple Synostosis Syndrome, Treacher Collins Syndrome, Treacher Collins-Franceschetti Syndrome 1, Trevor Disease, Triatrial Heart, Tricho-Dento-Osseous Syndrome, Trichodento Osseous Syndrome, Trichopoliodydystrophy, Trichorhinophalangeal Syndrome, Trichorhinophalangeal Syndrome, Tricuspid atresia, Trifunctional Protein
- 5 Deficiency, Trigeminal Neuralgia, Triglyceride Storage Disease Impaired Long-Chain Fatty Acid Oxidation, Trigonitis, Trigonocephaly, Trigonocephaly Syndrome, Trigonocephaly "C" Syndrome, Trimethylaminuria, Triphalangeal Thumbs-Hypoplastic Distal Phalanges-Onychodystrophy, Triphalangeal Thumb Syndrome, Triple Symptom Complex of Behcet, Triple X Syndrome, Triplo X Syndrome, Triploid Syndrome,
- 10 Triploidy, Triploidy Syndrome, Trismus-Pseudocamptodactyly Syndrome, Trisomy, Trisomy G Syndrome, Trisomy X, Trisomy 6q Partial, Trisomy 6q Syndrome Partial, Trisomy 9 Mosaic, Trisomy 9P Syndrome (Partial) Included, Trisomy 11q Partial, Trisomy 14 Mosaic, Trisomy 14 Mosaicism Syndrome, Trisomy 21 Syndrome, Trisomy 22 Mosaic, Trisomy 22 Mosaicism Syndrome, TRPS, TRPS1, TRPS2, TRPS3, True
- 15 Hermaphroditism, Truncus arteriosus, Tryptophan Malabsorption, Tryptophan Pyrrolase Deficiency, TS, TTP, TTTS, Tuberous Sclerosis, Tubular Ectasia, Turcot Syndrome, Turner Syndrome, Turner-Kieser Syndrome, Turner Phenotype with Normal Chromosomes (Karyotype), Turner-Varny Syndrome, Turricephaly, Twin-Twin Transfusion Syndrome, Twin-to-Twin Transfusion Syndrome, Type A, Type B, Type AB,
- 20 Type O, Type I Diabetes, Type I Familial Incomplete Male, Type I Familial Incomplete Male Pseudohermaphroditism, Type I Gaucher Disease, Type I (PCCA Deficiency), Type I Tyrosinemia, Type II Gaucher Disease, Type II Histiocytosis, Type II (PCCB Deficiency), Type II Tyrosinemia, Type IIA Distal Arthrogryposis Multiplex Congenita, Type III Gaucher Disease, Type III Tyrosinemia, Type III Dentinogenesis Imperfecta,
- 25 Typical Retinoschisis, Tyrosinase Negative Albinism (Type I), Tyrosinase Positive Albinism (Type II), Tyrosinemia type 1 acute form, Tyrosinemia type 1 chronic form, Tyrosinosis, UCE, Ulcerative Colitis, Ulcerative Colitis Chronic Non-Specific, Ulnar-Mammary Syndrome, Ulnar-Mammary Syndrome of Pallister, Ulnar Nerve Palsy, UMS, Unclassified FODs, Unconjugated Benign Bilirubinemia, Underactivity of Parathyroid,
- 30 Unilateral Ichthyosiform Erythroderma with Ipsilateral Malformations Limb, Unilateral Chondromatosis, Unilateral Defect of Pectoralis Muscle and Syndactyly of the Hand,

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- Unilateral Hemidysplasia Type, Unilateral Megalencephaly, Unilateral Partial Lipodystrophy, Unilateral Renal Agenesis, Unstable Colon, Unverricht Disease, Unverricht-Lundborg Disease, Unverricht-Lundborg-Laf Disease, Unverricht Syndrome, Upper Limb - Cardiovascular Syndrome (Holt-Oram), Upper Motor Neuron Disease,
- 5 Upper Airway Apnea, Urea Cycle Defects or Disorders, Urea Cycle Disorder Arginase Type, Urea Cycle Disorder Arginino Succinase Type, Urea Cycle Disorders Carbamyl Phosphate Synthetase Type, Urea Cycle Disorder Citrullinemia Type, Urea Cycle Disorders N-Acetyl Glutamate Synthetase Typ, Urea Cycle Disorder OTC Type, Urethral Syndrome, Urethro-Oculo-Articular Syndrome, Uridine Diphosphate
- 10 Glucuronosyltransferase Severe Def. Type I, Urinary Tract Defects, Urofacial Syndrome, Uroporphyrinogen III cosynthase, Urticaria pigmentosa, Usher Syndrome, Usher Type I, Usher Type II, Usher Type III, Usher Type IV, Uterine Synechiae, Uroporphyrinogen I-synthase, Uveitis, Uveomeningitis Syndrome, V-CJD, VACTERL Association, VACTERL Association, VACTERL Syndrome, Valgus Calcaneus, Valine Transaminase Deficiency,
- 15 Valinemia, Valproic Acid, Valproate acid exposure, Valproic acid exposure, Valproic acid, Van Buren's Disease, Van der Hoeve-Habertsma-Waardenburg-Gauldi Syndrome, Variable Onset Immunoglobulin Deficiency Dysgammaglobulinemia, Variant Creutzfeldt-Jakob Disease (V-CJD), Varicella Embryopathy, Variegate Porphyria, Vascular Birthmarks, Vascular Dementia Binswanger's Type, Vascular Erectile Tumor, Vascular
- 20 Hemophilia, Vascular Malformations, Vascular Malformations of the Brain, Vasculitis, Vasomotor Ataxia, Vasopressin-Resistant Diabetes Insipidus, Vasopressin-Sensitive Diabetes Insipidus, VATER Association, Vcf syndrome, Vcfs, Velocardiofacial Syndrome, VeloCardioFacial Syndrome, Venereal Arthritis, Venous Malformations, Ventricular Fibrillation, Ventricular Septal Defects, Congenital Ventricular Defects, Ventricular Septal
- 25 Defect, Ventricular Tachycardia, Venual Malformations, VEOHD, Vermis Aplasia, Vermis Cerebellar Agenesis, Vernal Keratoconjunctivitis, Verruca, Vertebral Anal Tracheoesophageal Esophageal Radial, Vertebral Ankylosing Hyperostosis, Very Early Onset Huntington's Disease, Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency, Vestibular Schwannoma, Vestibular Schwannoma Neurofibromatosis,
- 30 Vestibulocerebellar, Virchow's Oxycephaly, Visceral Xanthogranulomatosis, Visceral Xantho-Granulomatosis, Visceral myopathy-External Ophthalmoplegia, Visceromegaly-

- Umbilical Hernia-Macroglossia Syndrome, Visual Amnesia, Vitamin A Deficiency, Vitamin B-1 Deficiency, Vitelline Macular Dystrophy, Vitiligo, Vitiligo Capitis, Vitreoretinal Dystrophy, VKC, VKH Syndrome, VLCAD, Vogt Syndrome, Vogt Cephalosyndactyly, Vogt Koyanagi Harada Syndrome, Von Bechterew-Strumpell
- 5 Syndrome, Von Eulenburg Paramyotonia Congenita, Von Frey's Syndrome, Von Gierke Disease, Von Hippel-Lindau Syndrome, Von Mikulicz Syndrome, Von Recklinghausen Disease, Von Willebrandt Disease, VP, Vrolik Disease (Type II), VSD, Vulgaris Type Disorder of Cornification, Vulgaris Type Ichthyosis, W Syndrome, Waardenburg Syndrome, Waardenburg-Klein Syndrome, Waardenburg Syndrome Type I (WS1),
- 10 Waardenburg Syndrome Type II (WS2), Waardenburg Syndrome Type IIA (WS2A), Waardenburg Syndrome Type IIB (WS2B), Waardenburg Syndrome Type III (WS3), Waardenburg Syndrome Type IV (WS4), Waelsch's Syndrome, WAGR Complex, WAGR Syndrome, Waldenstroem's Macroglobulinemia, Waldenstrom's Purpura, Waldenstrom's Syndrome, Waldmann Disease, Walker-Warburg Syndrome, Wandering Spleen, Warburg
- 15 Syndrome, Warm Antibody Hemolytic Anemia, Warm Reacting Antibody Disease, Wartenberg Syndrome, WAS, Water on the Brain, Watson Syndrome, Watson-Alagille Syndrome, Waterhouse-Friderichsen syndrome, Waxy Disease, WBS, Weaver Syndrome, Weaver-Smith Syndrome, Weber-Cockayne Disease, Wegener's Granulomatosis, Weil Disease, Weil Syndrome, Weill-Marchesani, Weill-Marchesani Syndrome, Weill-Reyes
- 20 Syndrome, Weismann-Netter-Stuhl Syndrome, Weissenbacher-Zweymuller Syndrome, Wells Syndrome, Wenckebach, Werdnig-Hoffman Disease, Werdnig-Hoffman Paralysis, Werlhof's Disease, Werner Syndrome, Wernicke's (C) I Syndrome, Wernicke's aphasia, Wernicke-Korsakoff Syndrome, West Syndrome, Wet Beriberi, WHCR, Whipple's Disease, Whipple Disease, Whistling face syndrome, Whistling Face-Windmill Vane Hand
- 25 Syndrome, White-Darier Disease, Whitnall-Norman Syndrome, Whorled nevoid hypermelanosis, WHS, Wieacker Syndrome, Wieacher Syndrome, Wieacker-Wolff Syndrome, Wiedmann-Beckwith Syndrome, Wiedemann-Rautenstrauch Syndrome, Wildervanck Syndrome, Willebrand-Juergens Disease, Willi-Prader Syndrome, Williams Syndrome, Williams-Beuren Syndrome, Wilms' Tumor, Wilms' Tumor-Aniridia-
- 30 Gonadoblastoma-Mental Retardation Syndrome, Wilms Tumor Aniridia Gonadoblastoma Mental Retardation, Wilms' Tumor-Aniridia-Genitourinary Anomalies-Mental Retardation

Syndrome, Wilms Tumor-Pseudohermaphroditism-Nephropathy, Wilms Tumor and Pseudohermaphroditism, Wilms Tumor-Pseudohermaphroditism-Glomerulopathy, Wilson's Disease, Winchester Syndrome, Winchester-Grossman Syndrome, Wiskott-Aldrich Syndrome, Wiskott-Aldrich Type Immunodeficiency, Witkop Ectodermal
5 Dysplasias, Witkop Tooth-Nail Syndrome, Wittmaack-Ekbom Syndrome, WM Syndrome, WMS, WNS, Wohlfart-Disease, Wohlfart-Kugelberg-Welander Disease, Wolf Syndrome, Wolf-Hirschhorn Chromosome Region (WHCR), Wolf-Hirschhorn Syndrome, Wolff-Parkinson-White Syndrome, Wolfram Syndrome, Wolman Disease (Lysosomal Acid Lypase Deficiency), Woody Guthrie's Disease, WPW Syndrome, Writer's Cramp, WS, WSS,
10 WWS, Wyburn-Mason Syndrome, X-Linked Addison's Disease, X-linked Adrenoleukodystrophy (X-ALD), X-linked Adult Onset Spinobulbar Muscular Atrophy, X-linked Adult Spinal Muscular Atrophy, X-Linked Agammaglobulinemia with Growth Hormone Deficiency, X-Linked Agammaglobulinemia, Lymphoproliferate X-Linked Syndrome, X-linked Cardio myopathy and Neutropenia, X-Linked Centronuclear
15 myopathy, X-linked Copper Deficiency, X-linked Copper Malabsorption, X-Linked Dominant Conradi-Hunermann Syndrome, X-Linked Dominant Inheritance Agenesis of Corpus Callosum, X-Linked Dystonia-parkinsonism, X Linked Ichthyosis, X-Linked Infantile Agammaglobulinemia, X-Linked Infantile Nectrotizing Encephalopathy, X-linked Juvenile Retinoschisis, X-linked Lissencephaly, X-linked Lymphoproliferative
20 Syndrome, X-linked Mental Retardation-Clasped Thumb Syndrome, X-Linked Mental Retardation with Hypotonia, X-linked Mental Retardation and Macroorchidism, X-Linked Progressive Combined Variable Immunodeficiency, X-Linked Recessive Conradi-Hunermann Syndrome, X-Linked Recessive Severe Combined Immunodeficiency, X-Linked Retinoschisis, X-linked Spondyloepiphyseal Dysplasia, Xanthine Oxidase
25 Deficiency (Xanthinuria Deficiency, Hereditary), Xanthinuria Deficiency, Hereditary (Xanthine Oxidase Deficiency), Xanthogranulomatosis Generalized, Xanthoma Tuberosum, Xeroderma Pigmentosum, Xeroderma Pigmentosum Dominant Type, Xeroderma Pigmentosum Type A I XPA Classical Form, Xeroderma Pigmentosum Type B II XPB, Xeroderma Pigmentosum Type E V XPE, Xeroderma Pigmentosum Type C III
30 XPC, Xeroderma Pigmentosum Type D IV XPD, Xeroderma Pigmentosum Type F VI XPF, Xeroderma Pigmentosum Type G VII XPG, Xeroderma Pigmentosum Variant Type

XP-V, Xeroderma-Talipes-and Enamel Defect, Xerodermic Idiocy, Xerophthalmia, Xerotic Keratitis, XLP, XO Syndrome, XP, XX Male Syndrome, Sex Reversal, XXXXX Syndrome, XXY Syndrome, XYY Syndrome, XYY Chromosome Pattern, Yellow Mutant Albinism, Yellow Nail Syndrome, YKL, Young Female Arteritis, Yunis-Varon Syndrome,
5 YY Syndrome, Z-E Syndrome, Z- and -Protease Inhibitor Deficiency, Zellweger Syndrome, Zellweger cerebro-hepato-renal syndrome, ZES, Ziehen-Oppenheim Disease (Torsion Dystonia), Zimmermann-Laband Syndrome, Zinc Deficiency Congenital, Zinsser-Cole-Engman Syndrome, ZLS, Zollinger-Ellison Syndrome.

10 It is to be understood that unless otherwise indicated, the subject invention is not limited to specific formulations of components, manufacturing methods, dosage regimens or the like, as such may vary. It is also to be understood that the terminology used herein is for the purpose of describing particular embodiments only and is not intended to be limiting.

15 The singular forms "a", "an" and "the" include plural aspects unless the context clearly dictates otherwise. Thus, for example, reference to a PUFA includes reference to a single PUFA as well as two or more PUFAs or families of PUFAs, an agent includes a single agent, as well as two or more agents.

20 In describing and claiming the present invention, the following terminology is used in accordance with the definitions set forth below.

The terms "compound", "active agent", "chemical agent", "pharmacologically active agent", "medicament", "active" and "drug" are used interchangeably herein to refer to a
25 chemical compound that induces a desired pharmacological and/or physiological effect. All such terms also cover naturally occurring PUFAs and derivatives or modified forms thereof. The terms also encompass pharmaceutically acceptable and pharmacologically active ingredients of those active agents specifically mentioned herein including but not limited to salts, esters, amides, prodrugs, active metabolites, analogs and the like. When
30 the terms "compound", "active agent", "chemical agent" "pharmacologically active agent", "medicament", "active" and "drug" are used, then it is to be understood that this includes

the active agent *per se* as well as pharmaceutically acceptable, pharmacologically active salts, esters, amides, prodrugs, metabolites, analogs, etc.

Reference to a "compound", "active agent", "chemical agent" "pharmacologically active agent", "medicament", "active" or "drug" includes combinations of two or more actives such as two or more PUFAs or families of PUFAs. A "combination" also includes multi-part such as a two-part composition where the agents are provided separately and given or dispensed separately or admixed together prior to dispensation. For example, a multi-part pharmaceutical pack may have two or more agents separately maintained.

10

The term "combination" in addition, encompasses multivalent PUFAs such as two or more PUFAs linked *via* chemical bond formation.

In addition, the PUFAs may be co-administered with a range of other therapeutic agents including pain relievers such as opiates, preferably morphine, buprenorphine, levomethadone, codeine, tramadol or tilidine, non-steroidal analgesics, for example acetylsalicylic acid, paracetamol, diclofenac, meloxicam, ibuprofen, ibuprofen lysinate, ibuprofen in extruded form (as described in WO 99/06038), gabapentine or antidepressants, preferably imipramine, maprotiline, mianserine, fluoxetine, viloxazine, tranylcypromine and/or moclobemide.

The terms "effective amount" and "therapeutically effective amount" of an agent as used herein mean a sufficient amount of the agent (e.g. an agent such as a PUFA or a derivative thereof) to provide the desired therapeutic or physiological effect or outcome. Undesirable effects, e.g. side effects, are sometimes manifested along with the desired therapeutic effect; hence, a practitioner balances the potential benefits against the potential risks in determining what is an appropriate "effective amount". The exact amount required will vary from subject to subject, depending on the species, age and general condition of the subject, mode of administration and the like. Thus, it may not be possible to specify an exact "effective amount". However, an appropriate "effective amount" in any individual case may be determined by one of ordinary skill in the art using only routine

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experimentation.

By "pharmaceutically acceptable" carrier, excipient or diluent is meant a pharmaceutical vehicle comprised of a material that is not biologically or otherwise undesirable, i.e. the material may be administered to a subject along with the selected active agent without causing any or a substantial adverse reaction. Carriers may include excipients and other additives such as diluents, detergents, coloring agents, wetting or emulsifying agents, pH buffering agents, preservatives, and the like.

Similarly, a "pharmacologically acceptable" salt, ester, emide, prodrug or derivative of a compound as provided herein is a salt, ester, amide, prodrug or derivative that this not biologically or otherwise undesirable.

"Treating" a subject may involve prevention of a condition or other adverse physiological event in a susceptible individual as well as treatment of a clinically symptomatic individual by ameliorating the symptoms of the condition.

A "subject" as used herein refers to an animal, preferably a mammal and more preferably a human who can benefit from the pharmaceutical formulations and methods of the present invention. There is no limitation on the type of animal that could benefit from the presently described pharmaceutical formulations and methods. A subject regardless of whether a human or non-human animal may be referred to as an individual, patient, animal, host or recipient. The compounds and methods of the present invention have applications in human medicine, veterinary medicine as well as in general, domestic or wild animal husbandry. Non-human animals contemplated herein include livestock animals (e.g. sheep, pigs, cows, horses, donkeys), laboratory test animals (e.g. mice, rabbits, rats, guinea pigs), companion animals (e.g. dogs, cats) and captive wild animals.

The term "animals" include avian species such as poultry birds (e.g. chickens, ducks, turkeys, geese) and wild and game birds (e.g. wild ducks, pheasants, emus) and aviary birds.

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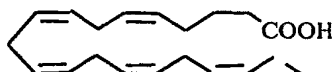
EXAMPLES

Examples 1

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Chemical Engineering of Fats

- Compounds were generated by the method described in WO 96/11908, WO 96/13507, WO 97/38688, WO 01/21172 and WO 01/21575 and are designated MP series, PT series and MP-PT hybrids. Molecules of the MP series possess the property of increased stability to oxidative breakdown. This reduced susceptibility to breakdown means that they are far less likely to cause the production of oxygen radicals which is the consequence of the metabolism of the natural omega-3 fatty acids. Molecules of the PT series also have this property but in addition are more soluble. The hybrid MP-PT series possess the above properties and demonstrate an expected outcome of higher antiinflammatory activity.
- 10 The structure of a natural fish oil fatty acid, ecosa pentaenoic acid, is shown in structure (a). The features of these types of fatty acids is a long carbon chain, unsaturation (double bonds) and a carboxyl Group (acid group) at one end of the chain.
- 15

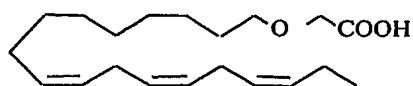


Fish oil fatty acid

(a)

- 20 The chemical engineering takes the form of *inter alia* substituting an oxygen atom (or sulphur) for the carbon, 2nd from the carboxyl group end (b). This is called the β -position. It is the area on the molecule on which the enzyme involved in the metabolism of the fats binds to. But because of the change the enzyme can not act on this group as efficiently as the unsubstituted molecule.. Thus the fat is handled differently by body tissues.
- 25 Advantage is taken of this.

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 β -oxa-21:3n-3

(b)

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Example 2**Treating Inflammatory Disease**

The naturally occurring ω -3 polyunsaturates (such as fish oil) have found use in the treatment of inflammatory diseases. These include the highly debilitating chronic forms
 10 such as rheumatoid arthritis, multiple sclerosis, inflammatory bowel disease and systemic lupus erythrocytosis. These are life-long diseases which are managed but cannot be cured. The principle mechanisms involve the T lymphocyte and macrophage and other white blood cells of the immune system (see Figure 1). These inappropriately attach to either joint tissue (in arthritis), blood vessel (in lupus), brain (multiple sclerosis) and gut tissue
 15 (inflammatory bowel disease) and then damage the tissue.

The PUFAs of the present invention target T-lymphocytes. When T-lymphocytes are exposed to MP5, for example, the cell takes up the fat as a nutritional requirement like any other fat but in this case the MP5 has a slight but vital change in its structure. MP5 stops
 20 the flow of a signal inside this cell preventing T-lymphocyte activation.

Example 3**Transplantation**

25 Management of patients with transplants involves the use of immunosuppressive medications e.g. cyclosporin which stops T lymphocyte activation. Rejection of transplanted tissues involves T-lymphocytes and macrophages in a similar manner to the delayed-type hypersensitivity (DTH) reaction. Thus MP5 has the potential to be used as

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a suitable immunosuppressive agent in transplantation especially because of the advantages it confers regarding safety compared to presently used immunosuppressants.

Example 4

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Treating Asthma and Allergy

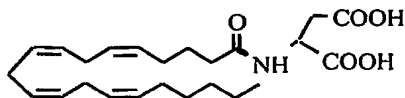
Tissues can be stimulated to produce fatty acid derived hormone like molecules called eicosanoids such as the leukotrienes . Production of these in an uncontrolled manner is known to lead to the appearance of serious diseases. These include asthma and allergic conditions. For example some leukotrienes act on the smooth muscle of the broncus of the airway preventing its relaxation leading to breathing difficulties as in asthma. In accordance with the present invention, a new form of polyunsaturates is provided as inhibitors of eicosanoid production and hence as potential medication to treat asthma and allergic conditions.

15

Example 5

Treating Pain

Some evidence has suggested that the novel fats may act on pathways involved in generating pain. As a consequence , some have been screened in two animal models of pain. The engineered polyunsaturates were found to act in a similar manner to aspirin but by a different pathway, giving these major advantages over toxicity problems associated with long term use of aspirin. One particular useful compound is PT2 (c). This is a polyunsaturated fatty acid which contains an amino acid covalently bound to its carboxyl group



20:4n-6 Asp (PT2)

(c)

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The chemical nature of these novel molecules suggests that they are easily delivered by skin application or oral administration. Investigations have demonstrated that after ingestion they soon appear in target organs (brain, kidney, lungs or skin). In preliminary studies in rats, active anti-inflammatory levels of these molecules do not display any toxic side effects. The significant anti-inflammatory property as well as the analgesic value of these molecules and their benign non-toxic nature makes the compounds ideal pharmaceuticals.

Example 6

Effects of Nitroanalog (Lx) of PUFA on PKC Activation

The effects of nitroanalogs of PUFAs on PKC activation were determined. Lx compounds at a concentration of 20 μ M were incubated with the HL-60 cell line (final condition 10^6 cells/ml) for 60 minutes. PKC activation was then attempted to be induced by PMA. PKC enzyme translocation was quantitated by Western blot. The results are shown in Table 1:

Table 1'- Inhibition of PKC Activation

PKC isozyme	Lx1	Lx2	Lx3	Lx4	Lx5	Lx6	Lx7	Lx8	Lx9
α	-	-	-	++	-	ND	+++	+++	-
$\beta 1$	+	-	-	+++	-	ND	+++	++	++
$\beta 2$	-	+++	+++	+++	-	ND	+++	+++	+++
δ	-	-	-	+++	-	ND	+	+++	+
ϵ	-	-	-	-	+	ND	+++	+++	+

+++ = strong inhibition of PKC activation, - = no inhibition of PKC activation, ND = not determined

It is evident that there are substantial differences in ability to inhibit the spectrum of five PKC isozymes by the different Lx compounds. For anti-cancer effect, δ and ϵ are of interest. These have been clearly associated with cell survival (ϵ) and cell death (δ). Take the example Lx7 and Lx8. Lx7 kills cancer cells very effectively but Lx8 poorly. The data in the Table shows that the activation of apoptotic protective isozyme ϵ is markedly inhibited by Lx7 without much inhibition of the activation of δ which promotes apoptosis.

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Therefore the cell dies. In contrast with Lx8 both isozymes are inhibited. The net effect is survival.

Although this argument is not conclusive with Lx9 where the compound is also strong in
5 killing cancer cells but there is balanced (+) inhibition of both δ and ϵ , there is a difference in other isozymes. This then decreases the activation of NF κ B which is required for survival in prostate cancer cells.

Example 7

10

Treatment of Systemic Vasculature

The aim of the experiment was to establish conditions for optimal activity of β -oxa 23:4n-6 (MP3) in relation to inhibition of up-regulation of adhesion molecular expression on the endothelium *in vivo* and to determine whether or not MP3 possesses anti-atherosclerotic
15 properties in experimental models.

It is proposed that β -oxa 23:4n-6 (MP3), through its ability to selectively inhibit the I κ B
kinase - NF κ B signalling pathway, inhibits the expression cell adhesion molecules on and the adherence of monocytes to the aortic endothelium, thus preventing the development of
20 atherosclerosis *in vivo*.

Atherosclerosis is a chronic inflammatory vascular disease which is characterised by a thickening of the vascular wall (atheroma) due to lipid accumulation and infiltration of circulating monocytes and T cells. The recruitment of monocytes into the intima in lesion
25 prone-sites is a key event in early atherogenesis. For this to occur, monocytes must first adhere to the endothelium at sites of endothelial injury or dysfunction caused by factors such as oxidised LDL, chylomicron remnants and/or advanced glycation end products (AGE) (Koya *et al*, *Diabetes* 47:859-866, 1998). Leukocyte adhesion to the endothelium and the subsequent emigration into the intima is mediated by leukocyte-endothelial cell
30 adhesion molecules (CAMs). These CAMs include the leukocyte L-selectin and the endothelial E-selectin, P-selectin, intercellular adhesion molecule (ICAM)-1 which binds